

# Combined burden and functional impact tests for cancer driver discovery using DriverPower

## Supplementary information

Shuai *et al.*, 2019

### Supplementary Note 1 Parameter tuning

As noted in the **Method** section, a single parameter  $F$  (the functional score threshold) was used to control the degree of functional adjustment in DriverPower. Functional scores used in DriverPower were positively correlated with the functional impact of mutations within elements. The parameter  $F$  controls the threshold score ( $S_T = -10\log_{10}F$ ) and must be within the interval  $(0,1]$ . The parameter  $F$  is negatively correlated with  $S_T$ . Genomic element with score  $S > S_T$  will be up-weighted; while element with score  $S < S_T$  will be down-weighted. Since DriverPower uses phred-scale scores,  $F$  can be interpreted as the proportion of functionally relevant variants among all observed variants. For instance,  $F = 0.01$  means  $S_T = 20$  so elements with functional score  $S > 20$  (i.e., rank top 1% in phred-like scale) will receive a functional weight  $>1$  and gain additional significance from functional impact information. Using larger  $F$  will result in smaller  $S_T$ , and more elements will obtain additional significance, we hence expect to obtain more driver candidates. Empirically speaking, larger  $F$  will cause higher recall but lower precision as illustrated in **Supplementary Fig. 16**.

The choice of  $F$  is dependent on the score scheme in use and/or the element set and tumour cohort in test. Here to avoid overfitting, we divided all 2583 donors into training donor set ( $N=1,117$ ) and test donor set ( $N=1,136$ ) to choose  $F$  for four score schemes (CADD, DANN, EIGEN and LINSIGHT) and three element types (CDS, splice site and other non-coding sites) separately. For CDS and splice site, we used the COSMIC cancer gene census (CGC) as the gold standard set for parameter tuning. For other non-coding sites, we used the PCAWG-raw as the gold standard set for parameter tuning. Precisions were calculated as the fraction of hits in the gold standard set and pseudo-recalls were calculated as the number of significant candidates with functional adjustment in gold standard over the number of nearly-significant candidates without functional adjustment in gold standard. This calculation of pseudo-recall measured the relative performance of DriverPower to its personal best. We observed that optimal parameters for the training donor set also worked in a similar way for the test donor set. Parameters learnt here were used for driver discovery in this analysis.

### Supplementary Note 2 Difference from the PCAWG freeze

The list of coding and non-coding drivers produced by this analysis (v1.0.0) differs slightly from the DriverPower results included in the PDFIG data analysis freeze due to the following reasons:

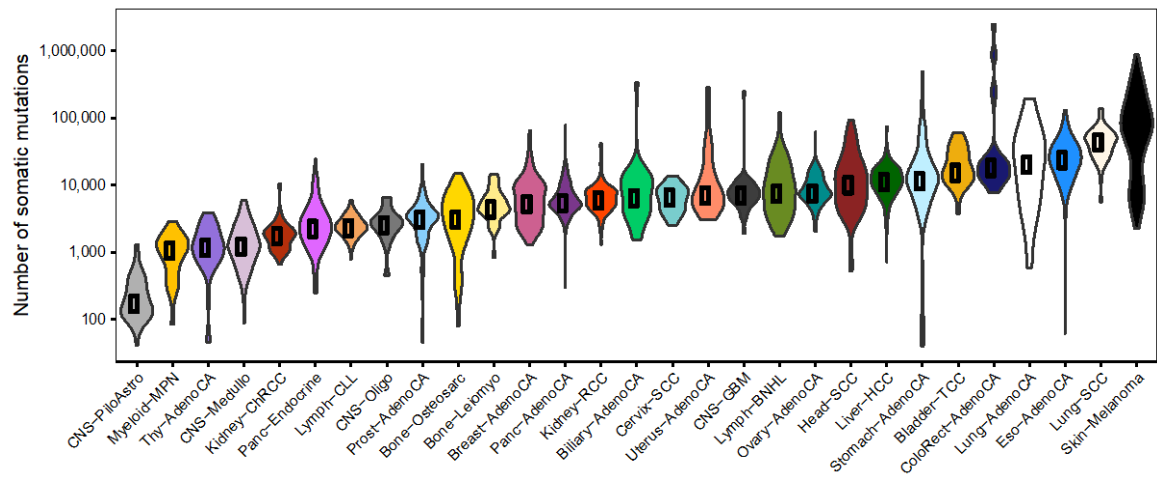
1. Hypermutated samples were not removed for the PCAWG freeze.
2. Only the binomial test was used in the PCAWG freeze.
3. The BMR model used in the PCAWG freeze was randomized lasso followed by GLM and in this analysis was gradient boosting machines.

4. Only CADD and EIGEN scores are used in the PCAWG freeze, and element p-value is the minimal p-value generated from two scores.

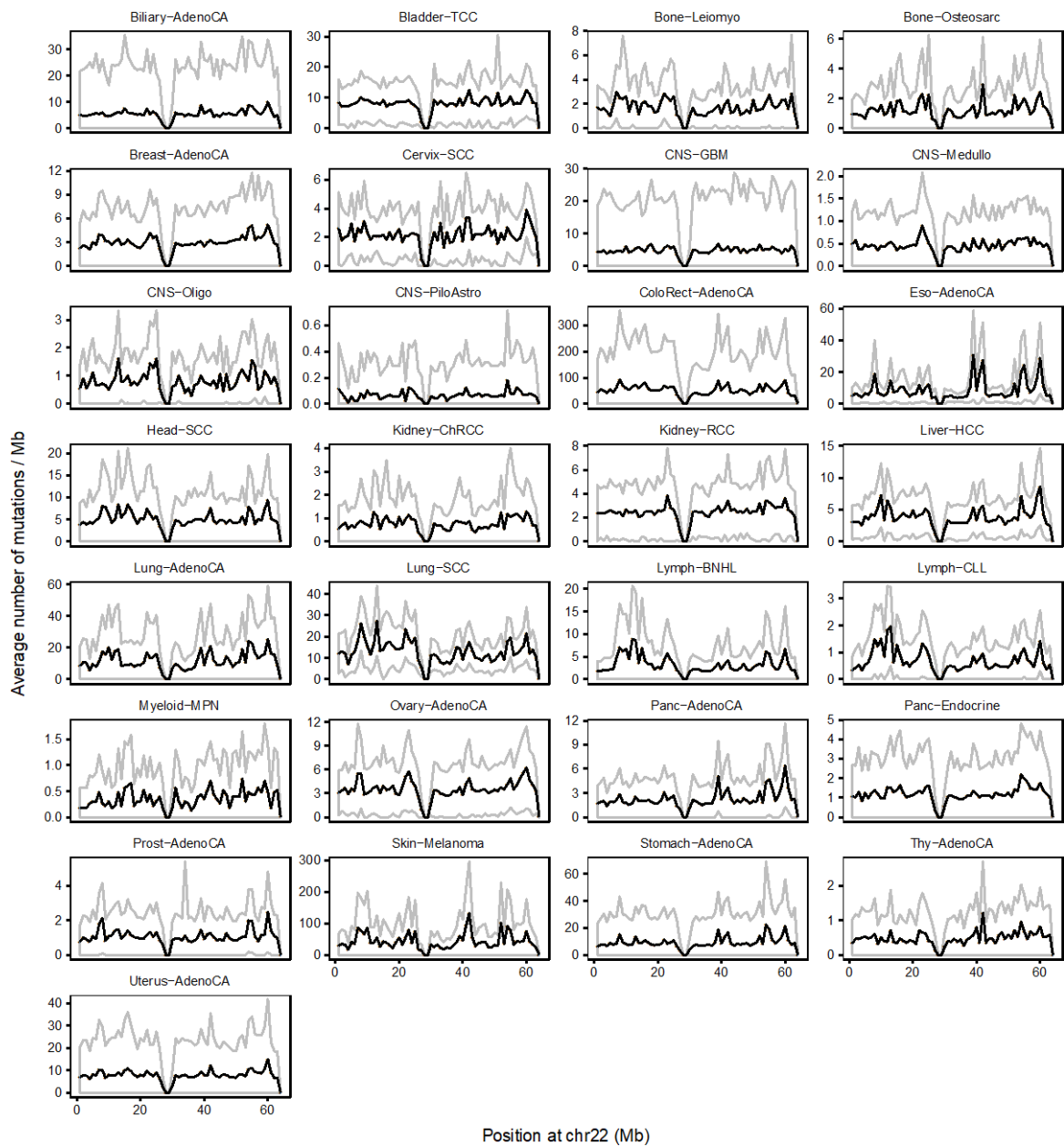
The threshold scores used for CADD and EIGEN in the PCAWG freeze were cohort-specific. Namely, 85%-95% percentile score of each tumour cohort was used as threshold.

Around 83% of candidates identified by v1.0.0 were also in PCAWG freeze (**Supplementary Fig. 17a,c**). For coding driver discovery, the current version has higher precision and lower recall (**Supplementary Fig. 17b**). For non-coding driver discovery, four candidates are only significant in the current version, including the promoter of lncRNA *RMRP* in Breast-AdenoCA and Stomach-AdenoCA, as well as *LEPROTL1* promoter in Bladder-TCC and *TERT* promoter in CNS-Oligo (**Supplementary Fig. 17d**). In addition, the removal of hypermutated samples and the incorporation of negative binomial test alleviated the inflation issue in melanomas and lymphomas.

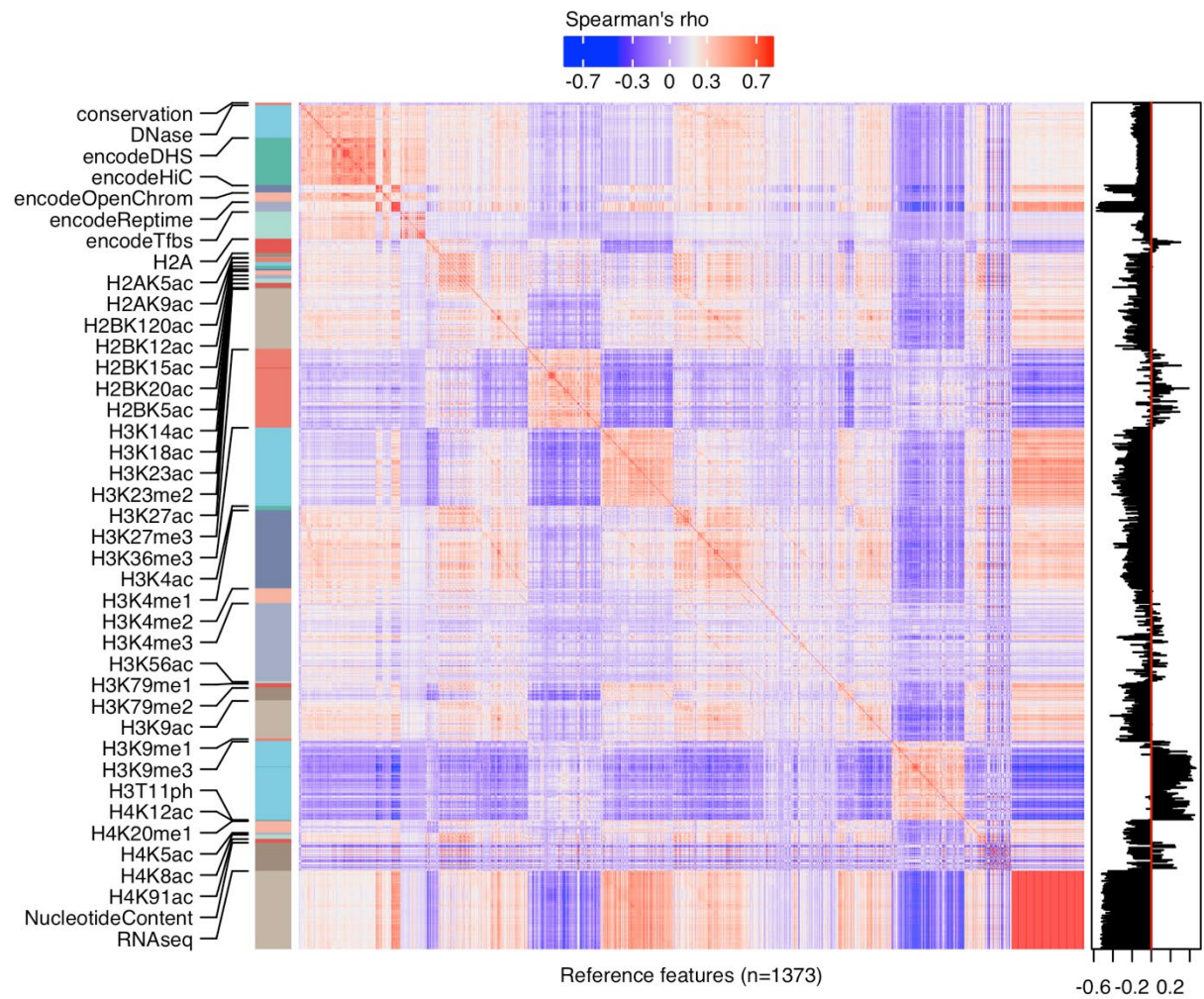
Supplementary Figure



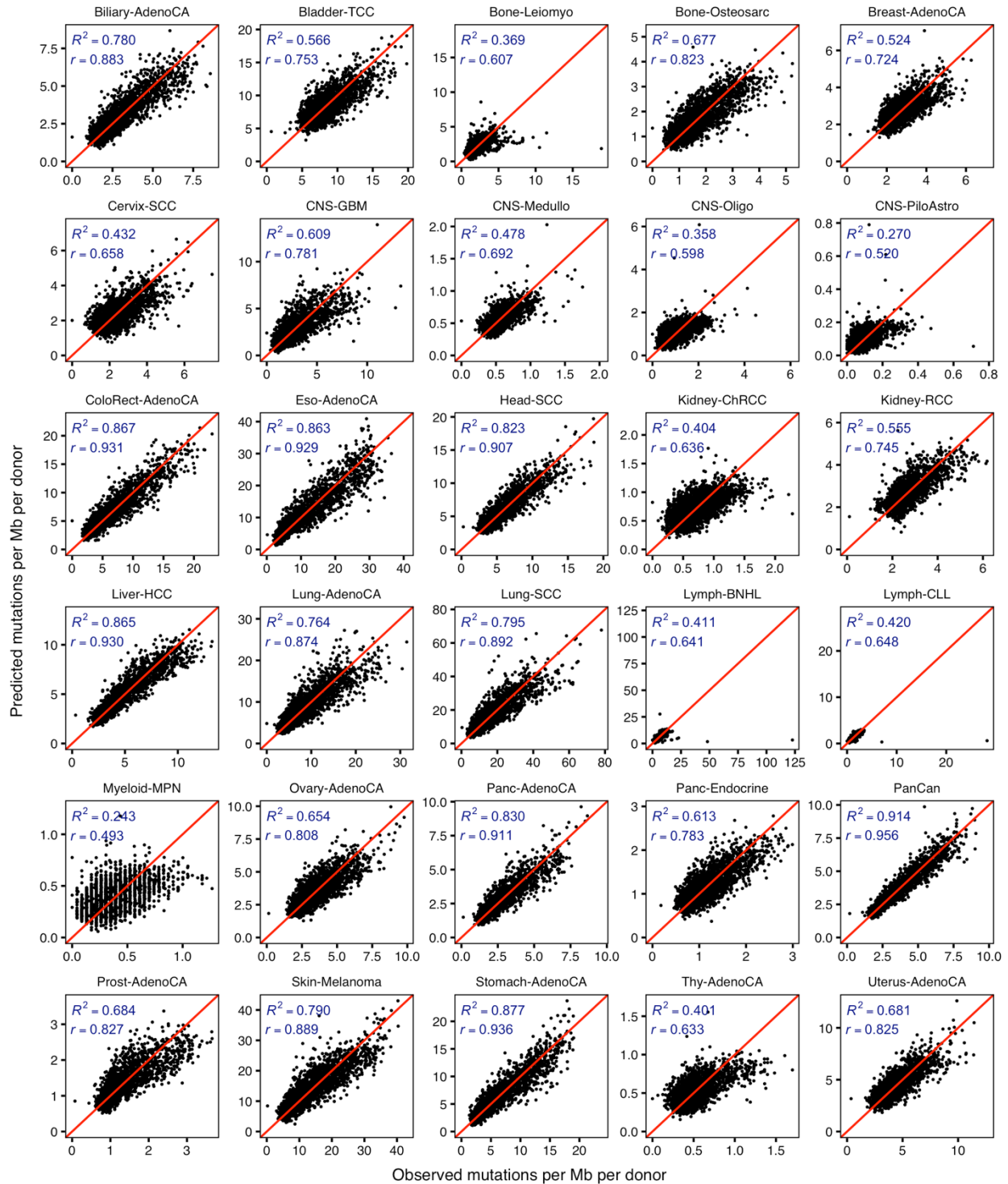
**Supplementary Figure 1 Heterogeneity at cohort and donor levels.** The violin plot shows the number of somatic mutations (log-scale) per donor across all tumour types. Hypermutated donors are included in the figure. Circles in the violin plot represent median values.



**Supplementary Figure 2 Heterogeneity at the locus level.** The average number of mutations per cohort per Mb are shown for chromosome 22. Black lines show the mean mutation rate per 1Mb interval. Filled ribbons show the mutation rate within 1 standard deviation.

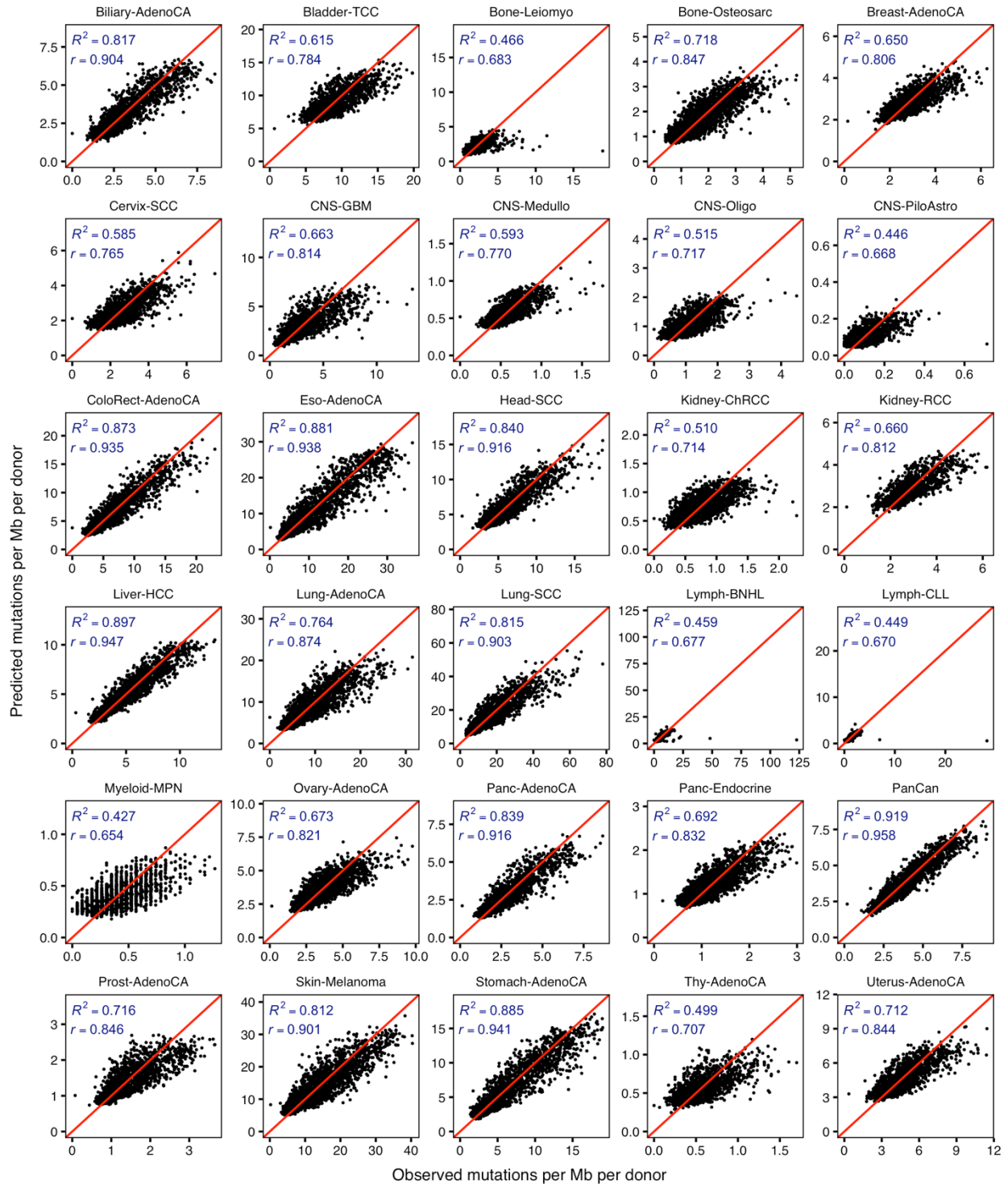


**Supplementary Figure 3 Features correlation matrix.** The heatmap shows the Spearman's rank correlation coefficient matrix between 1,373 features. Rows and columns are ordered by feature subgroups (40 subgroups in total). Barplot at right of the heatmap shows the Spearman's rho between features and mutation rates in training data for pan-cancer.

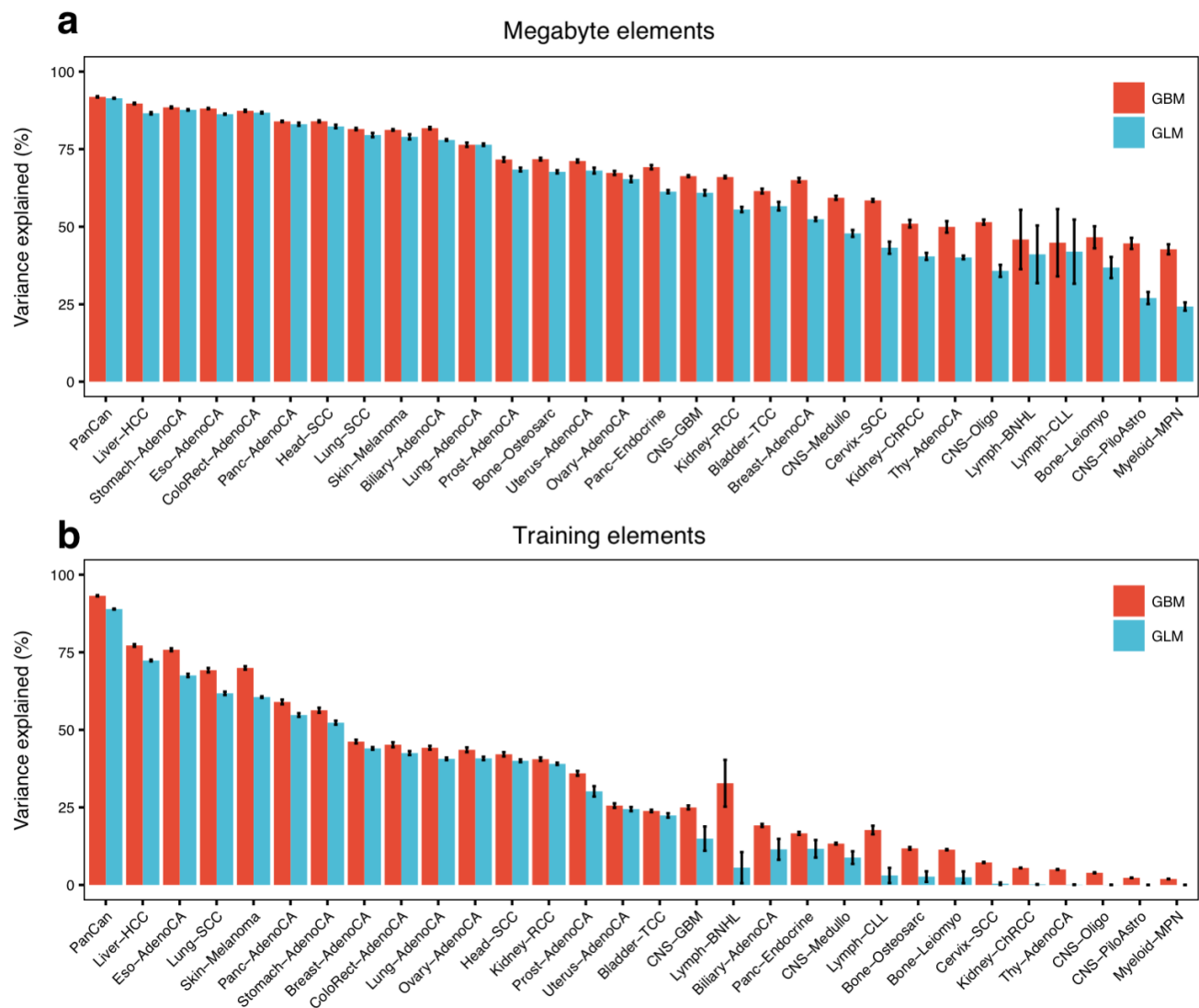


**Supplementary Figure 4 Expected versus GLM predicted mutation rate.** Mutation rates are the number of non-coding mutations within non-overlap 1Mb genome windows and normalized to per Mb per donor. Predictions are made from the randomized lasso and the generalized linear model. The  $R^2$  (coefficient of determination) and  $r$  (Pearson's correlation coefficients) are calculated with 5-fold cross validation. Note that only selected features are used in the GLM.



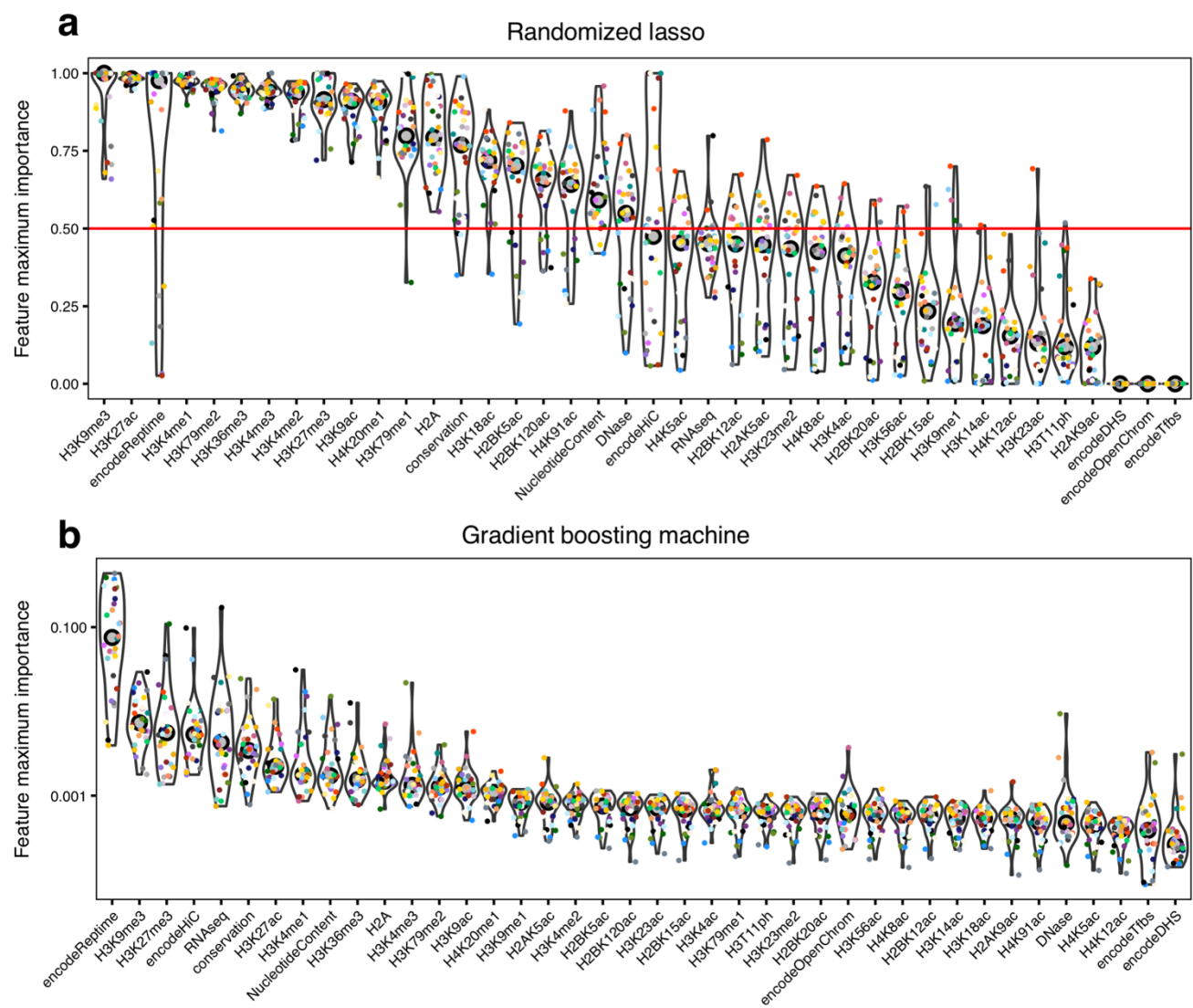


**Supplementary Figure 5 Expected versus GBM predicted mutation rate.** Mutation rates are the number of non-coding mutations within non-overlap 1Mb genome windows and normalized to per Mb per donor. Predictions are made from the gradient boosting machines. The  $R^2$  (coefficient of determination) and  $r$  (Pearson's correlation coefficients) are calculated with 5-fold cross validation. All features are used in the GBM.

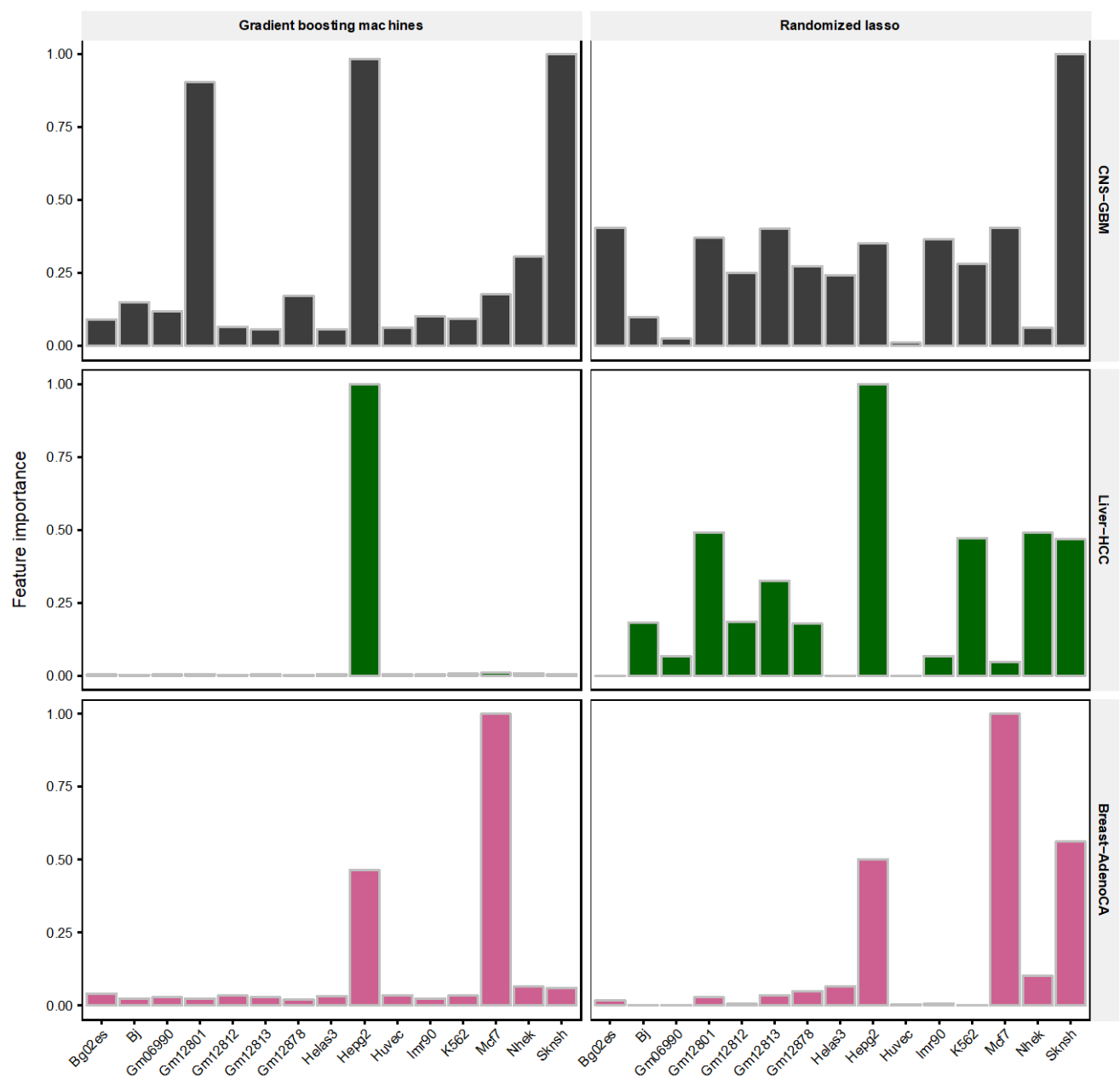


**Supplementary Figure 6 Model comparison.** (a) Variance explained for all tumour cohorts in 1Mb genome elements. (b) Variance explained for all tumour cohorts in the training element set. Error bars show the standard error of the mean (SEM). SEM is calculated from 5-fold cross validation. GBM is the gradient boosting machine. GLM is the generalized linear models.

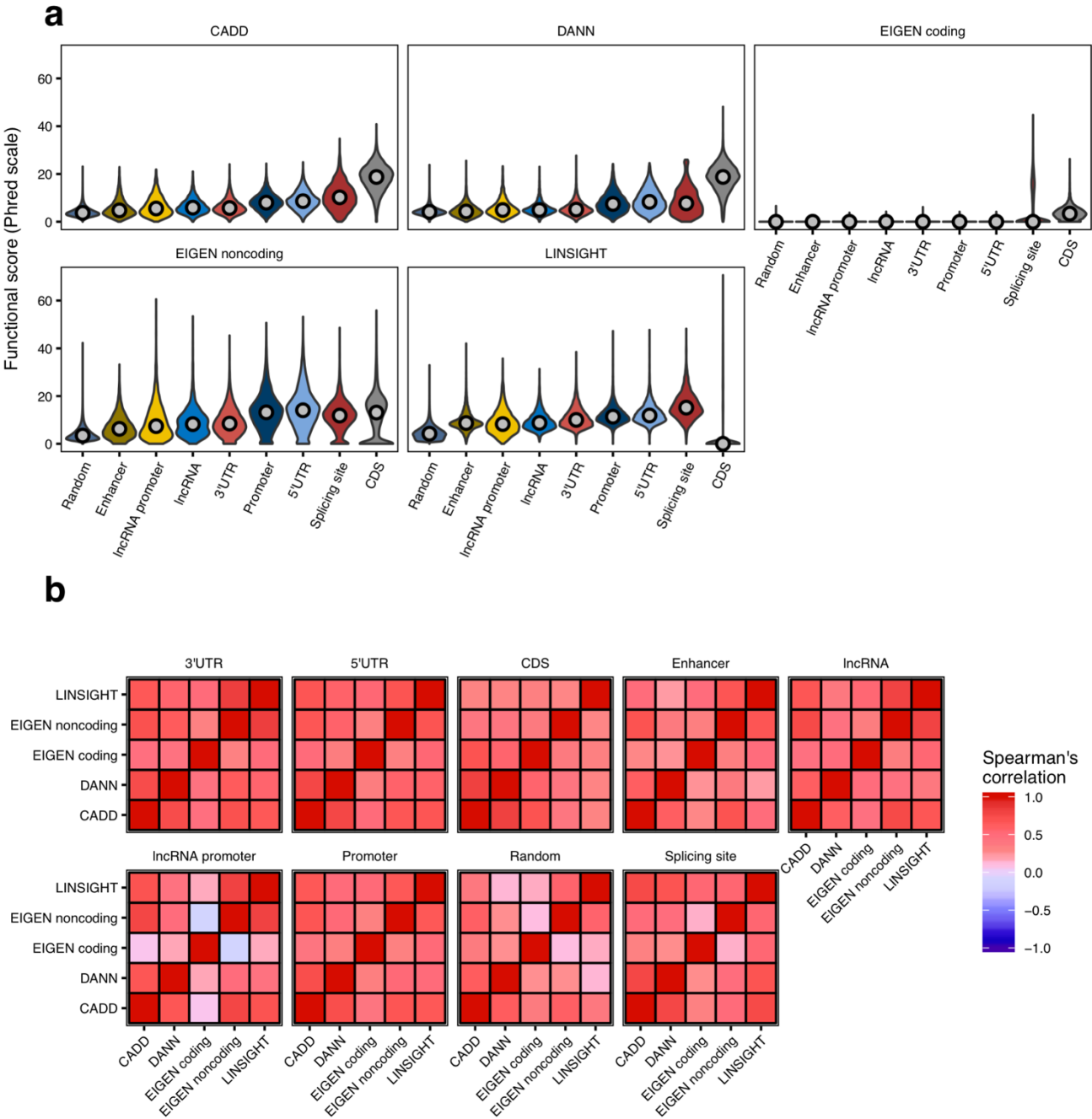




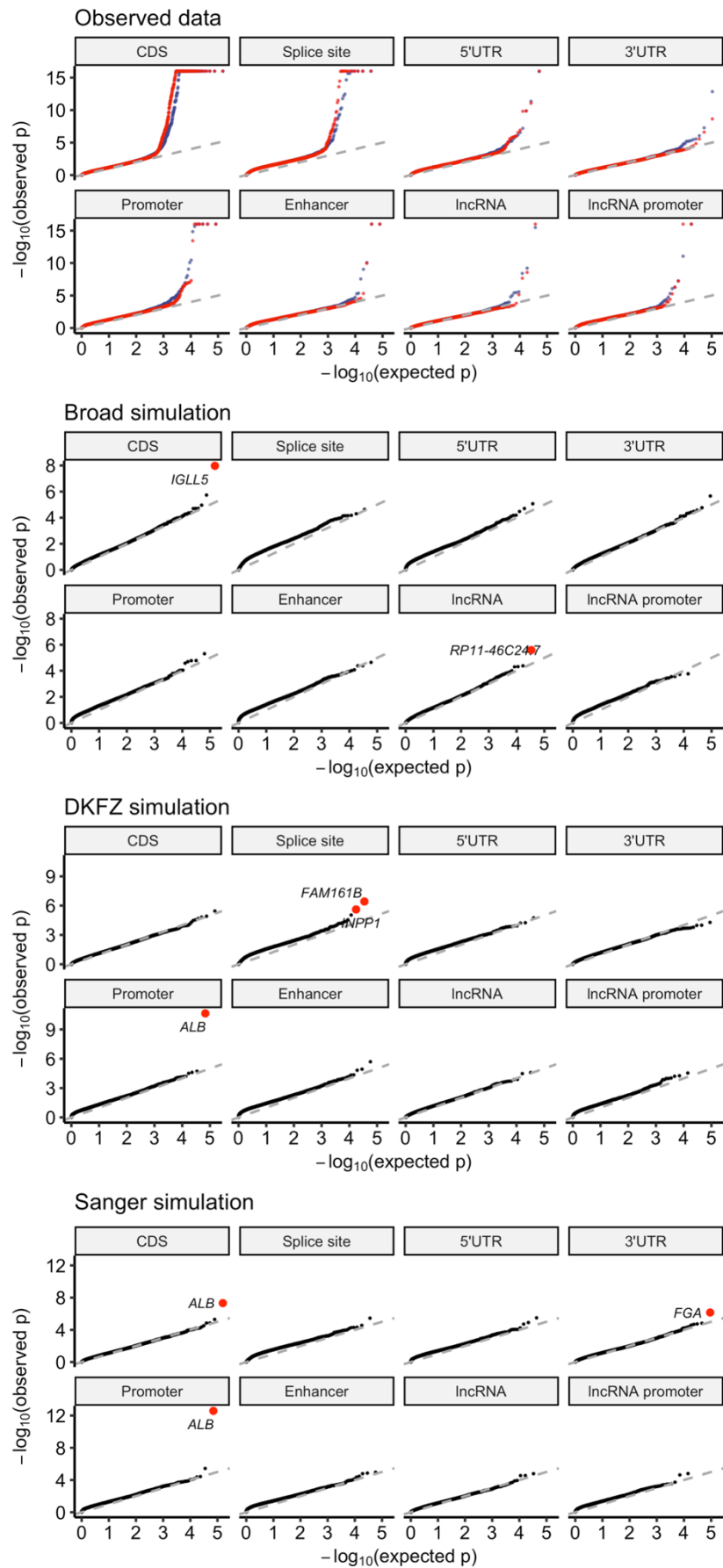
**Supplementary Figure 7 Feature importance rank.** All features are divided into subgroups (x-axis). Maximum importance of each group per tumour cohort (including pan-cancer) is represented as a point. Violin plot shows the distribution and median feature importance per subgroup among tumour cohorts. **(a)** Feature importance measures by the randomized lasso. Red line shows the 0.5 cutoff used in the generalized linear model. **(b)** Feature importance measures by the GBM (log-scale).



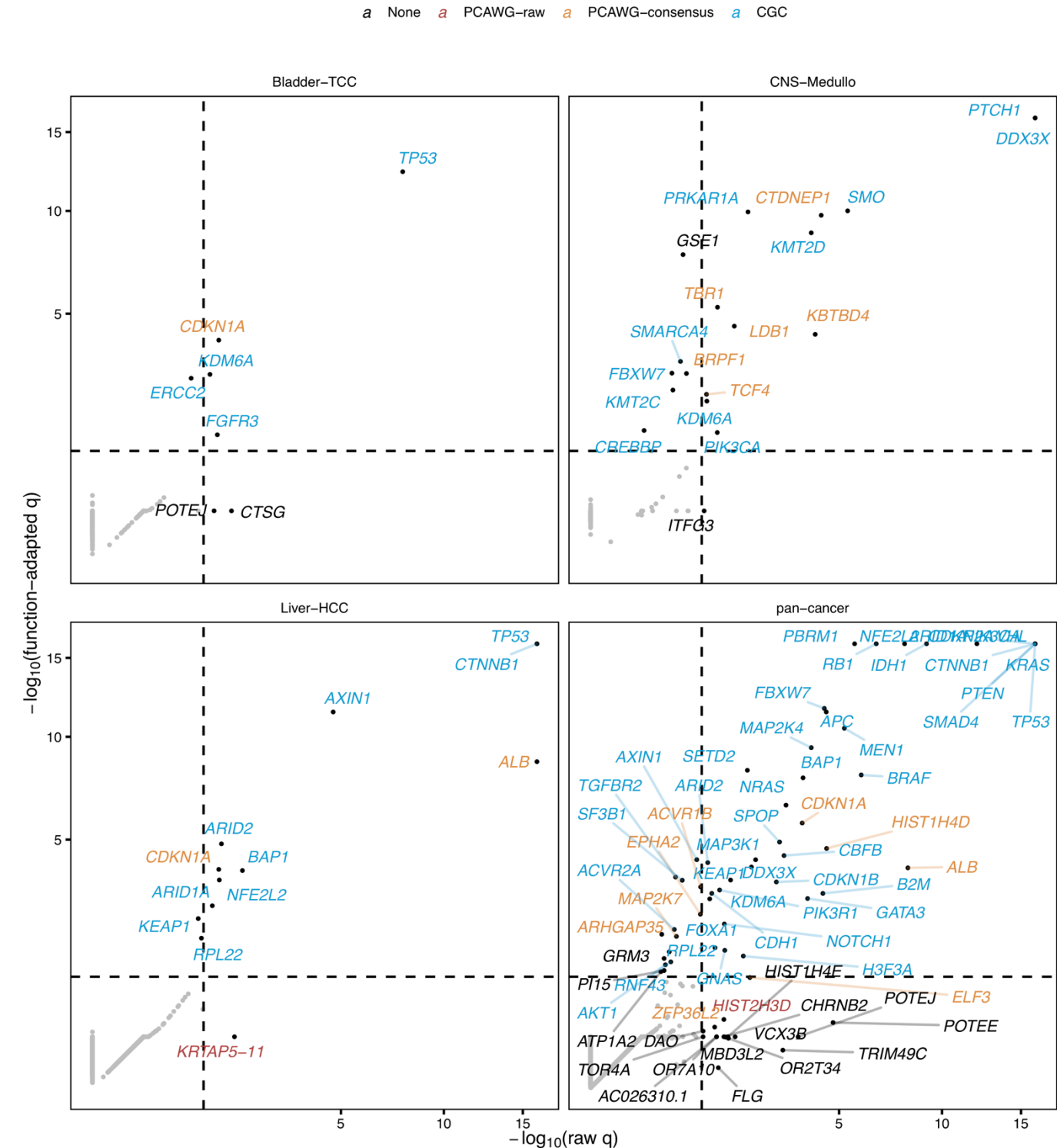
**Supplementary Figure 8 Top features associated with cell lines having similar origins.** Barplot shows the importance of 15 replication timing features for CNS-GBM, Liver-HCC and Breast-AdenoCA. X-axis shows the cell line name where the repli-seq experiment is conducted (ENCODE). Red line shows the 0.5 cutoff used in the feature selection. Feature weights from the GBM are normalized to [0, 1].



**Supplementary Figure 9 Elemental functional impact scores.** (a) Functional impact scores across all sets of elements for four different score schemes (CADD, DANN, EIGEN and LINSIGHT). Random elements are the training elements used in the model. (b) Spearman's correlation matrix for each set of elements (pairwise complete).



**Supplementary Figure 10 P-value quantile-quantile plots.** The p-value QQ-plots are shown for all test element types across three simulated datasets generated by the PDFIG and the observed PCAWG dataset. For the observed dataset, both raw p-values (blue) and function-adapted p-values (red) are shown. For the Broad, DKFZ and Sanger simulations, only raw p-values are shown and elements with  $q\text{-value} < 0.1$  are labelled. Each QQ-plot contains p-values from all tumour cohorts as the FDR control is performed in this way. Elements without mutations are removed in the plot. For better visualization,  $-\log_{10}$  observed p-values are capped at 16.

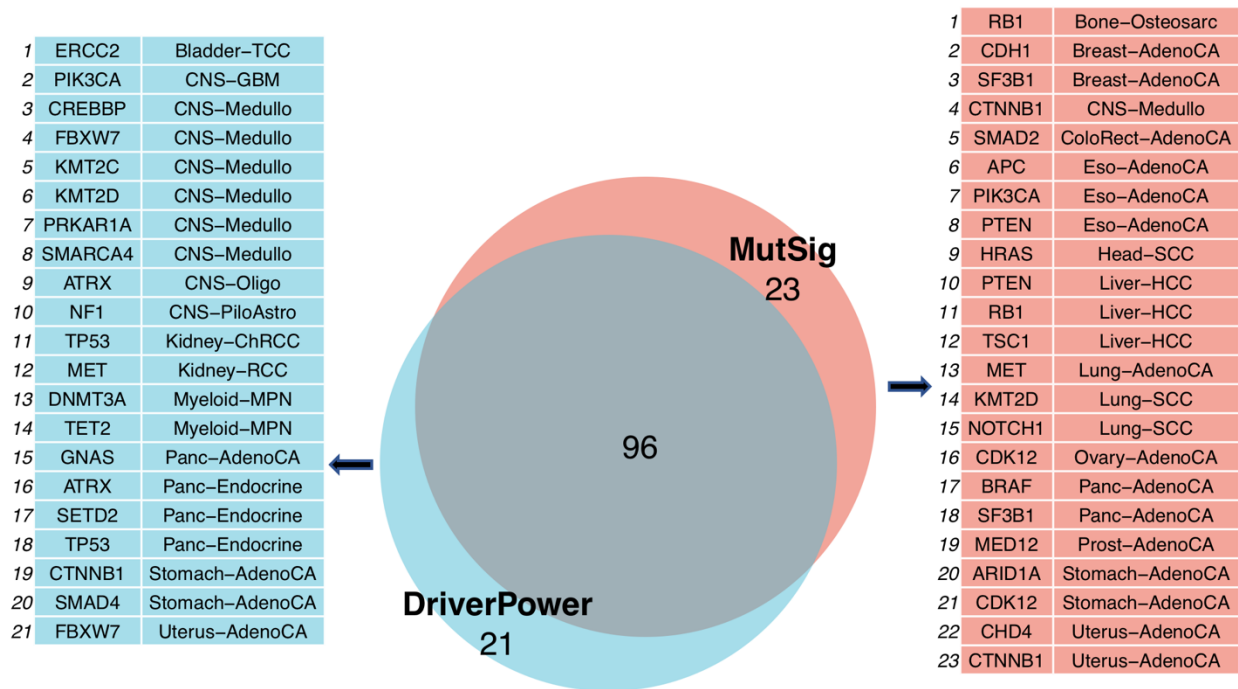


**Supplementary Figure 11 Functional adjustment improves CDS driver discovery.** Black horizontal and vertical dashed lines show the  $q$ -value = 0.1 cutoffs. Only significant genes are labelled with colors corresponding to reference gene sets. Function-adapted  $q$ -values are  $q$ -values with the usage of functional impact scores. Genes in the top-right quadrant are significant in both raw and functional adjusted results, in the bottom-left quadrant are significant in neither. Genes in the top-left

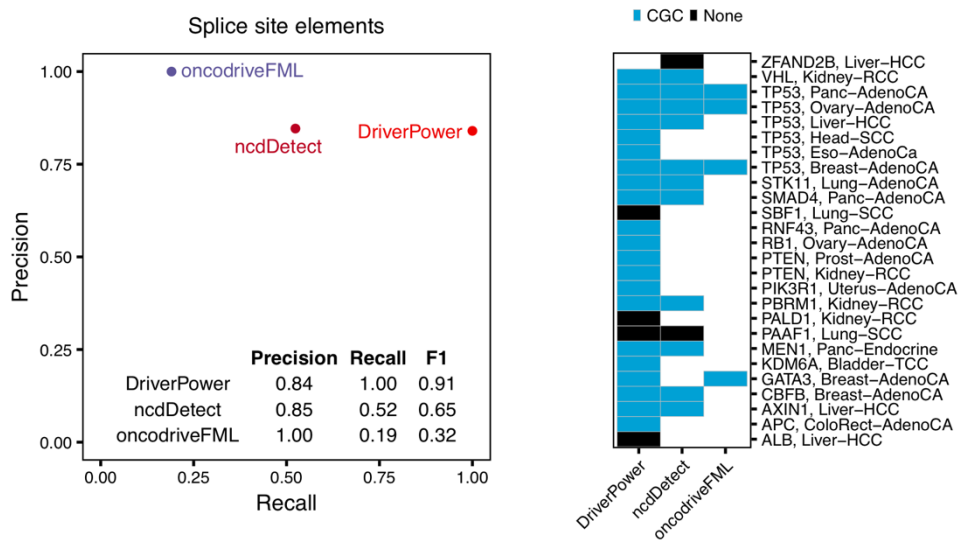


Supplementary Information (Shuai *et al.*, 2019)

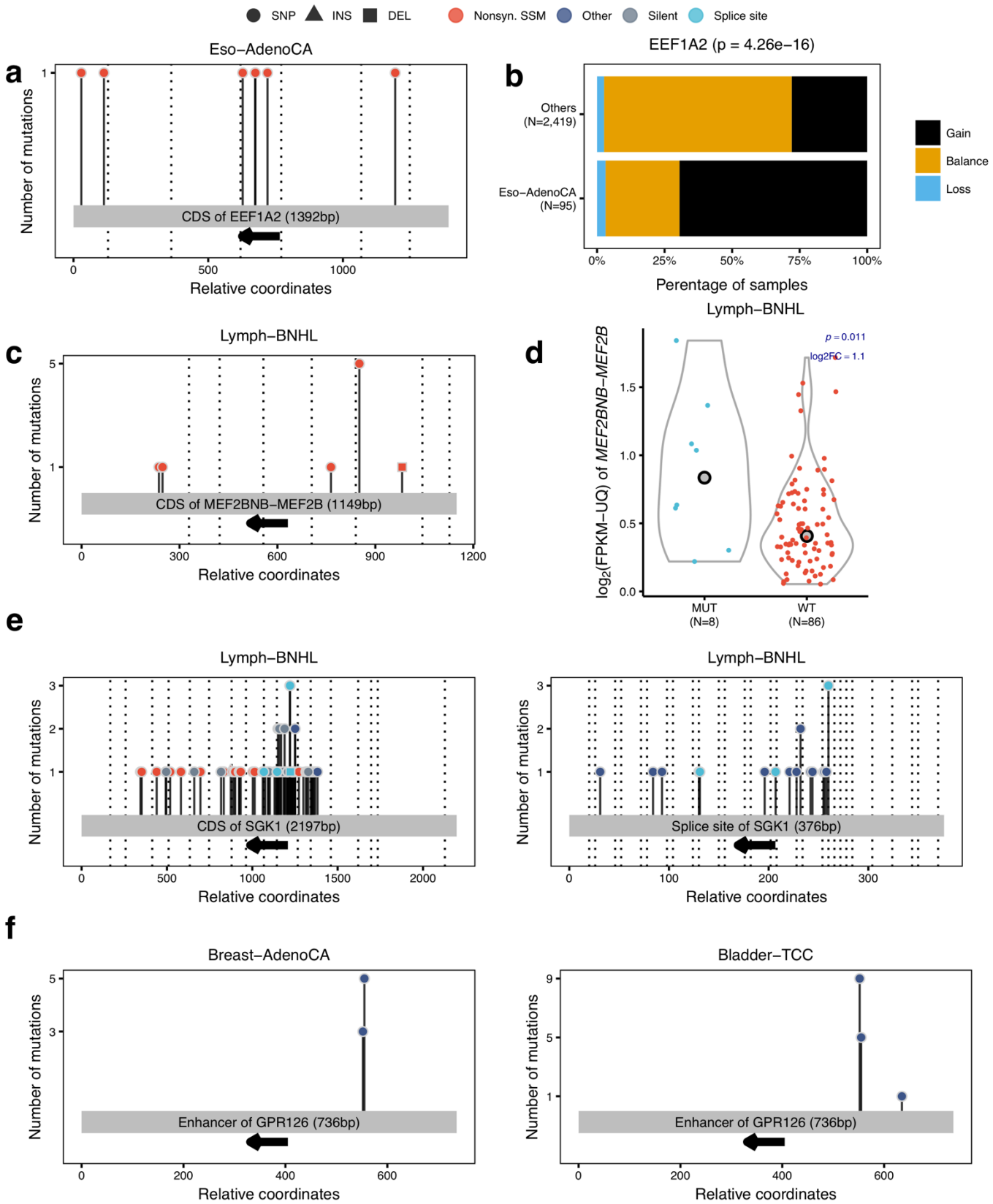
are significant only after functional adjustment and genes in the bottom-right are removed after functional adjustment.



**Supplementary Figure 12 Comparison of DriverPower and MutSig for CDS results.** The Venn diagram shows the relationship of true positive CDS results for DriverPower and MutSig. Significant genes present within the COSMIC CGC are considered to be true positive calls. DriverPower identifies 21 and MutSig identified 23 true positive genes exclusively.

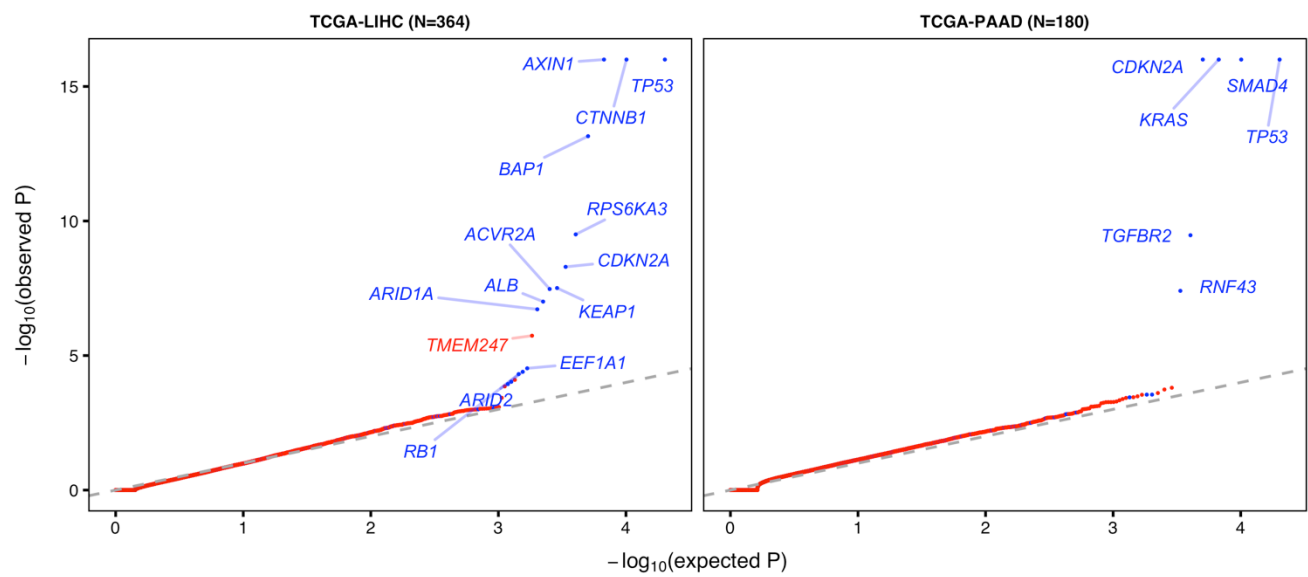


**Supplementary Figure 13 Benchmark result of splice sites.** The left panel shows the precision and recall for each method according to the results on 26 tumour cohorts. The right panel shows a heatmap of significant candidates identified by each method. Significant candidates in CGC are filled with blue and not in CGC are filled with black.



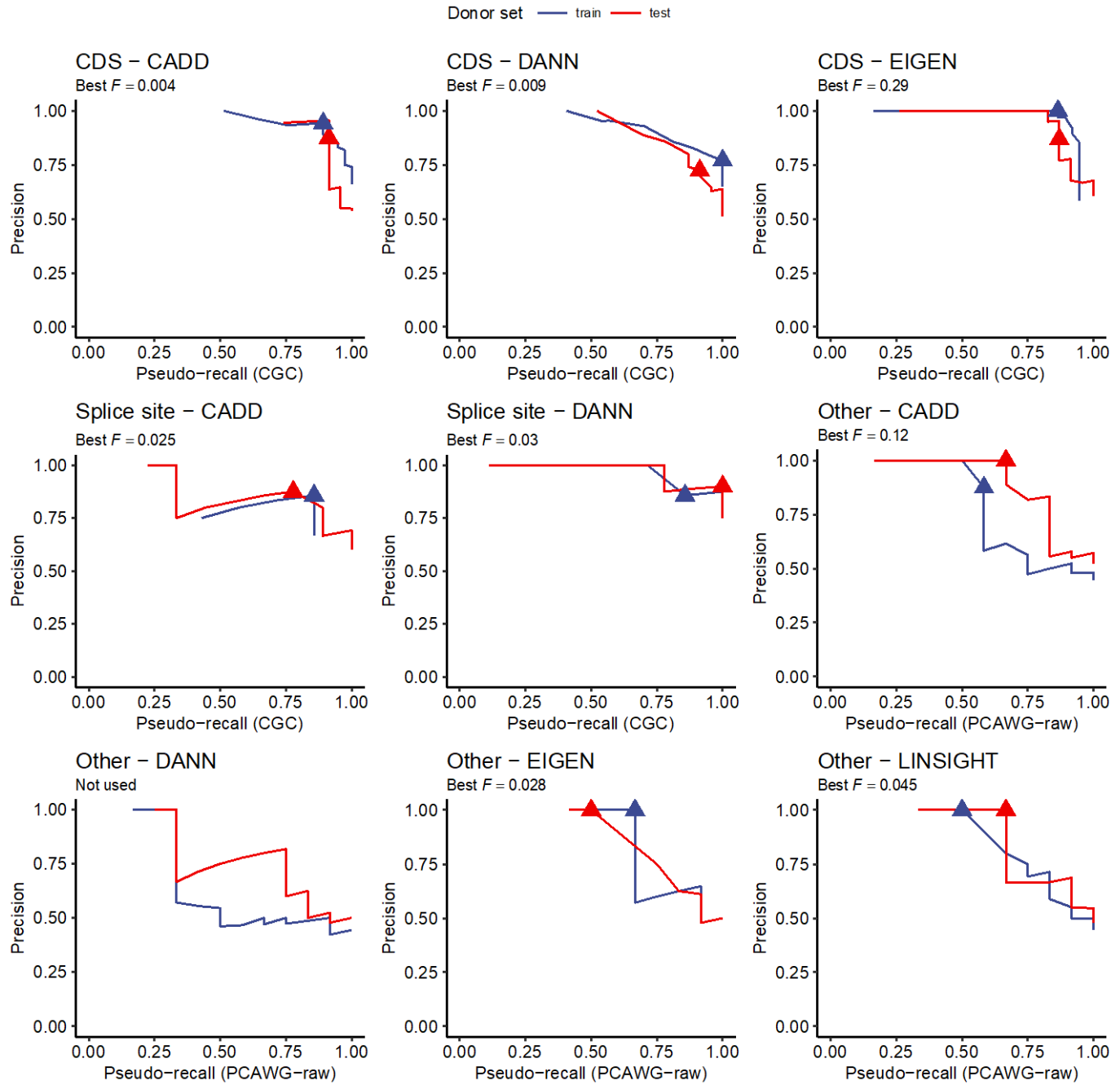
**Supplementary Figure 14 DriverPower-exclusive driver candidates overview.** (a,b) CDS of *EEF1A2* in Eso-AdenoCA. (a) is a mutation lollipop plot of *EEF1A2* in Eso-AdenoCA. (b) shows the

somatic copy number status for *EEF1A2* in Eso-AdenoCA or non-Eso-AdenoCA (other) samples; the p-value is from the Fisher's exact test. **(c)** A lollipop plot for CDS of *MEF2B* in Lymph-BNHL. **(d)** A violin plot for differential expression between Lymph-BNHL samples with mutated (MUT) *MEF2B* and non-mutated (WT) *MEF2B*. **(e)** Lollipop plots for CDS and splice site of *SGK1* in Lymph-BNHL. **(f)** Lollipop plots for *GPR126* enhancer in Breast-AdenoCA and Bladder-TCC. Lollipop plots show the distribution and classes of mutations (legends on top). In the lollipop plot, element is shown as a rectangular box and blocks of a disjoint element are separated by vertical dashed lines. Text within the box shows the name, type and length (bp) of elements. Arrow below the element box indicates the direction of element.

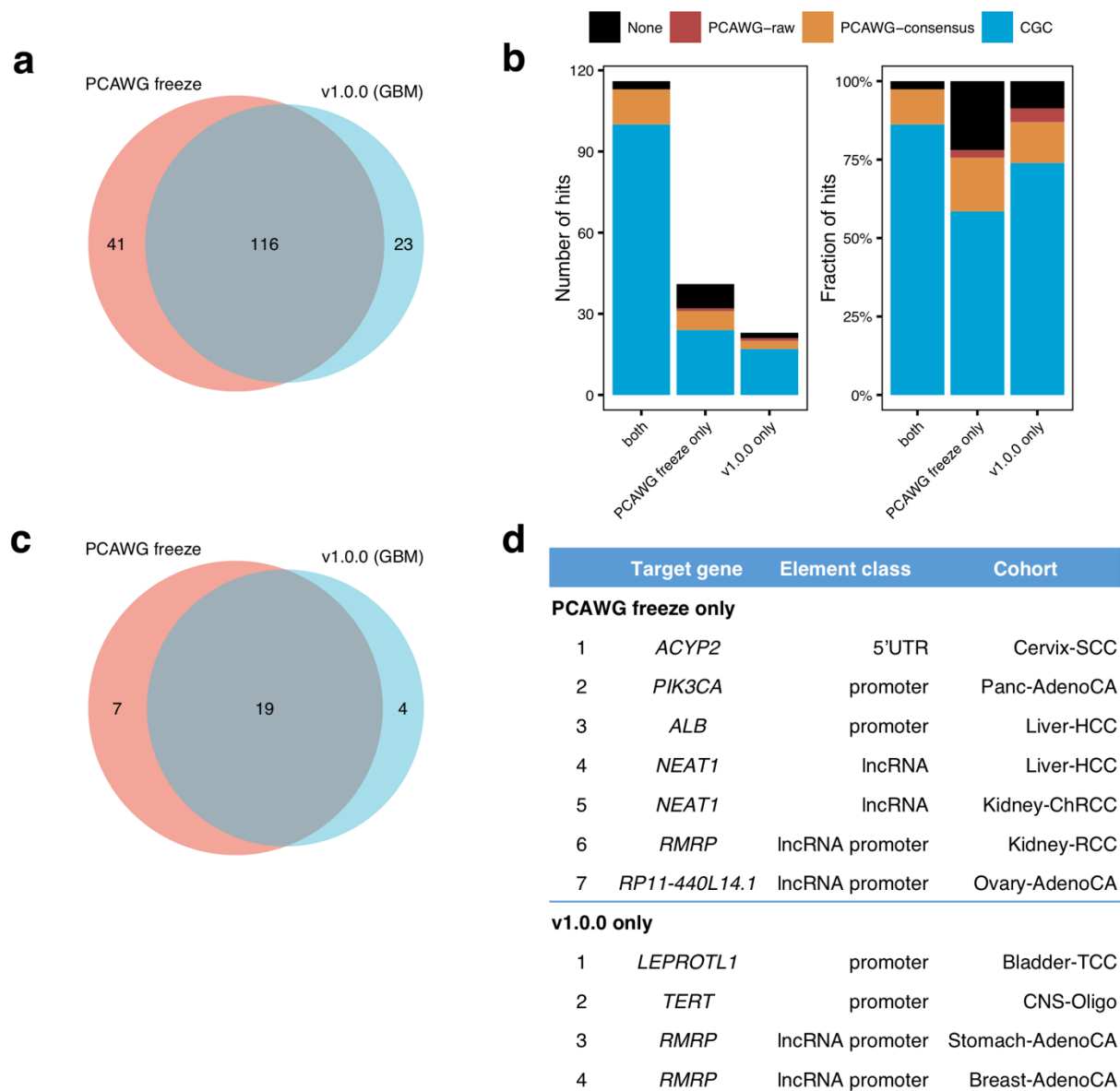


**Supplementary Figure 15 WES driver discovery result.** P-value Q-Q plots for two TCGA whole-exome sequencing datasets. Only significant genes are labelled ( $q < 0.1$ ). Blue points and labels indicate candidates within reference driver sets; red points and labels indicate likely false positive hits.





**Supplementary Figure 16 Parameterization of DriverPower.** Each panel shows a parameter ( $F$ ) search for one of the score schemes (CADD, DANN, EIGEN and LINSIGHT) and one of the element sets (CDS, splice site and other non-coding elements). The best  $F$  is indicated below each panel title. For each search path, the left end point is the smallest  $F$  searched and the right end point is the largest  $F$  searched; points correspond to  $F$  searched and triangles correspond to the best  $F$ . Search paths are colored for training (blue) and test (red) donor sets (see **Supplementary Note 1** for more details).



**Supplementary Figure 17 Comparison of PCAWG freeze and v1.0.0. (a,b)** Comparison of coding driver candidates for non-melanoma/lymphoma tumours. (a) is the venn plot showing the relationship between CDS results from PCAWG freeze and CDS results from this analysis (v1.0.0). (b) shows the number and fraction of candidates called for two versions. Columns in (b) are colored by reference gene sets. **(c,d)** Comparison of non-coding driver candidates for non-melanoma/lymphoma tumours. (c) is the venn plot showing the relationship of non-coding results (3'UTR, 5'UTR, promoter, enhancer, lncRNA and lncRNA promoter) between PCAWG freeze and v1.0.0. (d) shows all non-coding candidates that are unique to PCAWG freeze (n=7) or v1.0.0 (n=4). See **Supplementary Note 2** for more details.

**Supplementary Table1. Summary of datasets used in the paper.** Files are listed with a synapse ID (accession number), which refers to data organised on the Synapse platform (<https://www.synapse.org/>). In addition, every file has been mirrored at the International Cancer Genome Consortium Data Co-ordination Center (ICGC DCC), with the URL and filename reported in the table. The final column indicates whether the file is open access or is controlled tier, requiring authorisation to access it (instructions for access provided in the Data Availability statement and at <https://docs.icgc.org/pcawg/data/>). SNV, single nucleotide variant; MAF, Mutation Annotation Format (described at [https://docs.gdc.cancer.gov/Data/File\\_Formats/MAF\\_Format/](https://docs.gdc.cancer.gov/Data/File_Formats/MAF_Format/)); VCF, Variant Call Format (described at <https://www.internationalgenome.org/wiki/Analysis/vcf4.0/>).

Label	Synapse ID	ICGC DCC URL	ICGC DCC Filename	Access (Open/Controlled)
Consensus ICGC SNV+Indel	syn7364923	<a href="http://dcc.icgc.org/releases/PCAWG/consensus_snv_indel/">http://dcc.icgc.org/releases/PCAWG/consensus_snv_indel/</a>	final_consensus_snv_indel_passonly_icgc.public.tgz	Open
<b>Description:</b> The set of somatically acquired SNVs and indels across PCAWG tumour samples contributed by projects run under the auspices of ICGC. Variant calls were generated by three pipelines run independently on each sample, with subsequent merging into a consensus set of high-quality calls. The file is formatted using the MAF format.				
Consensus TCGA SNV+Indel	syn7364923	<a href="http://dcc.icgc.org/releases/PCAWG/consensus_snv_indel/">http://dcc.icgc.org/releases/PCAWG/consensus_snv_indel/</a>	final_consensus_passonly_snv_mnv_indel.tcga.controlled.maf.gz	Controlled
<b>Description:</b> The set of somatically acquired SNVs and indels across PCAWG tumour samples contributed by projects run under the auspices of TCGA. Variant calls were generated by three pipelines run independently on each sample, with subsequent merging into a consensus set of high-quality calls. The file is formatted using the MAF format.				
Gene expression profiles from RNA-Seq	syn5553991	<a href="http://dcc.icgc.org/releases/PCAWG/transcriptome/gene_expression/">http://dcc.icgc.org/releases/PCAWG/transcriptome/gene_expression/</a>	tophat_star_fpkm_uq.v2_allquot_gl.tsv.gz	Open
<b>Description:</b> .FPKM-UQ normalized gene expression profiles.				
Genomic interval lists	syn5259890	<a href="https://dcc.icgc.org/releases/PCAWG/drivers/metadata/genomic_intervals_lists">https://dcc.icgc.org/releases/PCAWG/drivers/metadata/genomic_intervals_lists</a>	Files with the name format [element_category].bed	Open
<b>Description:</b> Genomic elements used in driver calls. Files are in BED format.				
Driver region calls	syn7359546	<a href="https://dcc.icgc.org/releases/PCAWG/driver_mutations/significance_analysis/">https://dcc.icgc.org/releases/PCAWG/driver_mutations/significance_analysis/</a>		Controlled

**Description:** Driver candidates called by each individual methods using the same input data.

Gene-level CNV calls	syn8239175	<a href="https://dcc.icgc.org/releases/PCAWG/consensus_cnv/gene_level_calls">https://dcc.icgc.org/releases/PCAWG/consensus_cnv/gene_level_calls</a>	all_samples.consensus_level_calls.by_gene.170214.txt	Open
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**Description:** The set of somatically acquired copy number alterations across PCAWG tumour samples at the gene level. Codes: 0 is neutral, 1 is amplified, 2 is high-level amplified, -1 is deleted, -2 is deeply deleted.

Simulated mutations - Sanger	syn7187923	<a href="https://dcc.icgc.org/releases/PCAWG/drivers/metadata/simulated_mutations">https://dcc.icgc.org/releases/PCAWG/drivers/metadata/simulated_mutations</a>	PCAWG_randomised_sanger.icgc.controlled.tgz PCAWG_randomised_sanger.tcga.controlled.tgz	Controlled
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**Description:** A dataset of simulated mutations (Sanger).

Simulated mutations - DKFZ	syn7436065	<a href="https://dcc.icgc.org/releases/PCAWG/drivers/metadata/simulated_mutations">https://dcc.icgc.org/releases/PCAWG/drivers/metadata/simulated_mutations</a>	PCAWG_randomised_dkfz.icgc.controlled.tgz PCAWG_randomised_dkfz.tcga.controlled.tgz	Controlled
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**Description:** A dataset of simulated mutations (DKFZ).

Simulated mutations - Broad	syn7152699	<a href="https://dcc.icgc.org/releases/PCAWG/drivers/metadata/simulated_mutations">https://dcc.icgc.org/releases/PCAWG/drivers/metadata/simulated_mutations</a>	AugustRelease_Simulation_s_Broad.icgc.controlled.maf.gz AugustRelease_Simulation_s_Broad.tcga.controlled.maf.gz	Controlled
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**Description:** A dataset of simulated mutations (Broad).

Tumor histopathology	syn10389158	<a href="https://dcc.icgc.org/releases/PCAWG/clinical_and_histology">https://dcc.icgc.org/releases/PCAWG/clinical_and_histology</a>	pcawg_donor_clinical_August2016_v9.xlsx	Open
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**Description:** The tumour subtypes were hand-curated and harmonised to icd-0-3 organ system and histological descriptions using a semi-automated process, and then grouped into a series of tiers using a tumour subtype grouping system. This grouping system was reviewed and approved by a group of pathology experts under the coordination of Dr. David Louis at Massachusetts General Hospital. The format is a spreadsheet.

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### Annotations working group

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### Quality control working group

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Matthew H Bailey<sup>138,139</sup>, Beifang Niu<sup>140</sup>, Matthias Bieg<sup>81,141</sup>, Paul C Boutros<sup>9,133,142,143</sup>, Ivo Buchhalter<sup>52,53,54</sup>, Adam P Butler<sup>2</sup>, Ken Chen<sup>144</sup>, Zechen Chong<sup>145</sup>, **Li Ding**<sup>#138,139,146</sup>, Oliver Drechsel<sup>136,147</sup>, Lewis Jonathan Dursi<sup>9,65</sup>, Roland Eils<sup>52,54,66,67</sup>, Kyle Ellrott<sup>51</sup>, Shadrielle MG Espiritu<sup>9</sup>, Yu Fan<sup>148</sup>, Robert S Fulton<sup>138,139,146</sup>, Shengjie Gao<sup>149</sup>, Josep Ll Gelpi<sup>46,71</sup>, Mark Gerstein<sup>120,121,122,123</sup>, Gad Getz<sup>3,4,5,6</sup>, Santiago Gonzalez<sup>7,8</sup>, Ivo G Gut<sup>135,136</sup>, Faraz Hach<sup>150,151</sup>, Michael C Heinold<sup>52,54</sup>, Julian M Hess<sup>3,75</sup>, Jonathan Hinton<sup>2</sup>, Taobo Hu<sup>152</sup>, Vincent Huang<sup>9</sup>, Yi Huang<sup>153,154</sup>, Barbara Hutter<sup>80,81,82</sup>, David R Jones<sup>2</sup>, Jongsun Jung<sup>89</sup>, Natalie Jäger<sup>52</sup>, Hyung-Lae Kim<sup>27</sup>, Kortine Kleinheinz<sup>52,54</sup>, Sushant Kumar<sup>122,123</sup>, Yogesh Kumar<sup>152</sup>, Christopher M Lalansingh<sup>9</sup>, Ignaty Leshchiner<sup>3</sup>, Ivica Letunic<sup>155</sup>, Dimitri Livitz<sup>3</sup>, Eric Z Ma<sup>152</sup>, Yosef E Maruvka<sup>3,75,106</sup>, R Jay Mashl<sup>139,156</sup>, Michael D McLellan<sup>138,139,146</sup>, Andrew Menzies<sup>2</sup>, Ana Milovanovic<sup>46</sup>, Morten Muhlig Nielsen<sup>125</sup>, Stephan Ossowski<sup>136,147,157</sup>, Nagarajan Paramasivam<sup>52,81</sup>, Jakob Skou Pedersen<sup>125,132</sup>, Marc D Perry<sup>45,103</sup>, Montserrat Puiggròs<sup>105</sup>, Keiran M Raine<sup>2</sup>, Esther Rheinbay<sup>3,6,106</sup>, Romina Royo<sup>105</sup>, S Cenk Sahinalp<sup>151,158,159</sup>, Gordon Saksena<sup>3</sup>, Iman Sarrafi<sup>151,159</sup>, Matthias Schlesner<sup>52,110</sup>, **Jared T Simpson**<sup>#9,160</sup>, Lucy Stebbings<sup>2</sup>, Chip Stewart<sup>3</sup>, Miranda D Stobbe<sup>135,136</sup>, Jon W Teague<sup>2</sup>, Grace Tiao<sup>3</sup>, David Torrents<sup>105,111</sup>, Jeremiah A Wala<sup>3,6,49</sup>, Jiayin Wang<sup>139,154,161</sup>, Wenyi Wang<sup>148</sup>, Sebastian M Waszak<sup>8</sup>, Joachim Weischenfeldt<sup>8,113,114</sup>, Michael C Wendl<sup>139,162,163</sup>, Johannes Werner<sup>52,115</sup>, David A Wheeler<sup>164,165</sup>, Zhenggang Wu<sup>152</sup>, Hong Xue<sup>152</sup>, Sergei Yakneen<sup>8</sup>, Takafumi N Yamaguchi<sup>9</sup>, Kai Ye<sup>161,166</sup>, Venkata D Yellapantula<sup>167,168</sup>, Christina K Yung<sup>45</sup> and Junjun Zhang<sup>45</sup>

### Drivers and functional interpretation



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## Integration of transcriptome and genome

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Hiroiyuki Aburatani<sup>272</sup>, **Benjamin P Berman**<sup>#273,274,275</sup>, Hans Binder<sup>276,277</sup>, **Benedikt Brors**<sup>#82,119,278</sup>, Huy Q Dinh<sup>273</sup>, Lars Feuerbach<sup>119</sup>, Shengjie Gao<sup>149</sup>, Ivo G Gut<sup>135,136</sup>, Simon C Heath<sup>135,136</sup>, Steve Hoffmann<sup>277,279,280,281</sup>, Charles D Imbusch<sup>119</sup>, Ekta Khurana<sup>117,118,130,131</sup>, Helene Kretzmer<sup>277,281</sup>, Peter W Laird<sup>282</sup>, Jose I Martin-Subero<sup>111,283</sup>, Genta Nagae<sup>272,284</sup>, **Christoph Plass**<sup>#285</sup>, Paz Polak<sup>3,4,6</sup>, Hui Shen<sup>286</sup>, Reiner Siebert<sup>262,761</sup>, Nasa Sinnott-Armstrong<sup>3,61</sup>, Miranda D Stobbe<sup>135,136</sup>, Qi Wang<sup>98</sup>, Dieter Weichenhan<sup>285</sup>, Sergei Yakneen<sup>8</sup> and Wanding Zhou<sup>286</sup>

## Patterns of structural variations, signatures, genomic correlations, retrotransposons, mobile elements

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Ludmil B Alexandrov<sup>2,317</sup>, Erik N Bergstrom<sup>318</sup>, Arnoud Boot<sup>267,319</sup>, Paul C Boutros<sup>9,133,142,143</sup>, Kin Chan<sup>293</sup>, Kyle Covington<sup>165</sup>, Akihiro Fujimoto<sup>48</sup>, Gad Getz<sup>3,4,5,6</sup>, Dmitry A Gordenin<sup>297</sup>, Nicholas J Haradhvala<sup>3,106</sup>, Mi Ni Huang<sup>267,319</sup>, S. M. Ashiqul Islam<sup>317</sup>, Marat D Kazanov<sup>303,304,305</sup>, Jaegil Kim<sup>3</sup>, Leszek J Klimczak<sup>306</sup>, Michael S Lawrence<sup>3,47,106</sup>, Iñigo Martincorena<sup>2</sup>, John R McPherson<sup>267,319</sup>, Sandro Morganello<sup>2</sup>, Ville Mustonen<sup>320,321,322</sup>, Hidewaki Nakagawa<sup>48</sup>, Avlin Wei Tian Ng<sup>323</sup>, Serena Nik-Zainal<sup>2,324,325,326</sup>, Paz Polak<sup>3,4,6</sup>, Stephenie D Prokopec<sup>9</sup>, Steven A Roberts<sup>313</sup>, **Steven G Rozen**<sup>#267,268,319</sup>, Radhakrishnan Sabarinathan<sup>183,190,214</sup>, Natalie Saini<sup>297</sup>, Tatsuhiro Shibata<sup>33,34</sup>, Yuichi Shiraishi<sup>87</sup>, **Michael Rudolf Stratton**<sup>#2</sup>, **Bin Tean Teh**<sup>#266,267,268,269,270</sup>, Ignacio Vázquez-García<sup>2,167,327,328</sup>, Yang Wu<sup>267,319</sup>, Fouad Yousif<sup>9</sup> and Willie Yu<sup>329</sup>

## Germline cancer genome

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## Tumor subtypes and clinical translation

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## Evolution and heterogeneity

David J Adams<sup>2</sup>, Pavana Anur<sup>373</sup>, Rameen Beroukhim<sup>3,6,172</sup>, Paul C Boutros<sup>9,133,142,143</sup>, David D L Bowtell<sup>187,291</sup>, Peter J Campbell<sup>1,2</sup>, Shaolong Cao<sup>148</sup>, Elizabeth L Christie<sup>187</sup>, Marek Cmero<sup>374,375,376</sup>, Yupeng Cun<sup>377</sup>, Kevin J Dawson<sup>2</sup>, Jonas Demeulemeester<sup>63,64</sup>, Stefan C Dentre<sup>2,64,354</sup>, Amit G Deshwar<sup>378</sup>, Nilgun Donmez<sup>151,159</sup>, Ruben M Drews<sup>294</sup>, Roland Eils<sup>52,54,66,67</sup>, Yu Fan<sup>148</sup>, Matthew W Fittall<sup>64</sup>, Dale W Garsed<sup>187,188</sup>, Moritz Gerstung<sup>7,8</sup>, Gad Getz<sup>3,4,5,6</sup>, Santiago Gonzalez<sup>7,8</sup>, Gavin Ha<sup>3</sup>, Kerstin Haase<sup>64</sup>, Marcin

Imielinski<sup>299,300</sup>, Lara Jerman<sup>8,379</sup>, Yuan Ji<sup>380,381</sup>, Clemency Jolly<sup>64</sup>, Kortine Kleinheinz<sup>52,54</sup>, Juhee Lee<sup>382</sup>, Henry Lee-Six<sup>2</sup>, Ignaty Leshchiner<sup>3</sup>, Dimitri Livitz<sup>3</sup>, Geoff Macintyre<sup>294</sup>, Salem Malikic<sup>151,159</sup>, Florian Markowetz<sup>294,295</sup>, Iñigo Martincorena<sup>2</sup>, Thomas J Mitchell<sup>2,295,383</sup>, Quaid D Morris<sup>358,384</sup>, Ville Mustonen<sup>320,321,322</sup>, Layla Oesper<sup>385</sup>, Martin Peifer<sup>377</sup>, Myron Peto<sup>386</sup>, Benjamin J Raphael<sup>120</sup>, Daniel Rosebrock<sup>3</sup>, Yulia Rubanova<sup>160,358</sup>, S Cenk Sahinalp<sup>151,158,159</sup>, Adriana Salcedo<sup>9</sup>, Matthias Schlesner<sup>52,110</sup>, Steven E Schumacher<sup>3,217</sup>, Subhajit Sengupta<sup>387</sup>, Ruian Shi<sup>384</sup>, Seung Jun Shin<sup>264</sup>, **Paul T Spellman**<sup>#388</sup>, Oliver Spiro<sup>3</sup>, Lincoln D Stein<sup>9,10</sup>, Maxime Tarabichi<sup>2,64</sup>, **Peter Van Loo**<sup>#63,64</sup>, Shankar Vembu<sup>384,389</sup>, Ignacio Vázquez-García<sup>2,167,327,328</sup>, Wenyi Wang<sup>148</sup>, **David C Wedge**<sup>#2,354,355</sup>, David A Wheeler<sup>164,165</sup>, Jeffrey A Wintersinger<sup>192,358,390</sup>, Tsun-Po Yang<sup>377</sup>, Xiaotong Yao<sup>299,316</sup>, Kaixian Yu<sup>391</sup>, Ke Yuan<sup>294,369,372</sup> and Hongtu Zhu<sup>392,393</sup>

### Exploratory: portals, visualization and software infrastructure

Fatima Al-Shahrour<sup>357</sup>, Elisabet Barrera<sup>7</sup>, Wojciech Bazant<sup>7</sup>, Alvis Brazma<sup>7</sup>, Isidro Cortés-Ciriano<sup>237,238,239</sup>, Brian Craft<sup>240</sup>, David Craft<sup>3</sup>, Vincent Ferretti<sup>45,69</sup>, Nuno A Fonseca<sup>7,70</sup>, Anja Füllgrabe<sup>7</sup>, Mary J Goldman<sup>240</sup>, **David Haussler**<sup>#240,394</sup>, Wolfgang Huber<sup>8</sup>, Maria Keays<sup>7</sup>, Alfonso Muñoz<sup>7</sup>, Brian D O'Connor<sup>45,50</sup>, Irene Papatheodorou<sup>7</sup>, Robert Petryszak<sup>7</sup>, Elena Piñeiro-Yáñez<sup>357</sup>, Alfonso Valencia<sup>105,111</sup>, **Miguel Vazquez**<sup>#105,112</sup>, John N Weinstein<sup>395,396</sup>, Qian Xiang<sup>116</sup>, Junjun Zhang<sup>45</sup> and **Jingchun Zhu**<sup>#240</sup>

### Exploratory: mitochondrial variants and HLA/immunogenicity

Peter J Campbell<sup>1,2</sup>, Yiwen Chen<sup>148</sup>, Chad J Creighton<sup>241</sup>, Li Ding<sup>138,139,146</sup>, Akihiro Fujimoto<sup>48</sup>, Masashi Fujita<sup>48</sup>, Gad Getz<sup>3,4,5,6</sup>, Leng Han<sup>231</sup>, Takanori Hasegawa<sup>87</sup>, Shuto Hayashi<sup>87</sup>, Seiya Imoto<sup>86,87</sup>, Young Seok Ju<sup>2,181</sup>, Hyung-Lae Kim<sup>27</sup>, Youngwook Kim<sup>96,97</sup>, Youngil Koh<sup>307,308</sup>, Mitsuhiro Komura<sup>87</sup>, Jun Li<sup>148</sup>, **Han Liang**<sup>#397</sup>, Iñigo Martincorena<sup>2</sup>, Satoru Miyano<sup>87</sup>, Shinichi Mizuno<sup>398</sup>, **Hidewaki Nakagawa**<sup>#48</sup>, Keunchil Park<sup>206,207</sup>, Eigo Shimizu<sup>87</sup>, Yumeng Wang<sup>148,399</sup>, John N Weinstein<sup>395,396</sup>, Yanxun Xu<sup>400</sup>, Rui Yamaguchi<sup>87</sup>, Fan Yang<sup>384</sup>, Yang Yang<sup>231</sup>, Christopher J Yoon<sup>181</sup>, Sung-Soo Yoon<sup>308</sup>, Yuan Yuan<sup>148</sup>, Fan Zhang<sup>246</sup> and Zemin Zhang<sup>246,271</sup>

### Exploratory: pathogens

Malik Alawi<sup>401,402</sup>, Ivan Borožan<sup>9</sup>, Daniel S Brewer<sup>403,404</sup>, Colin S Cooper<sup>404,405,406</sup>, Nikita Desai<sup>45</sup>, **Roland Eils**<sup>#52,54,66,67</sup>, Vincent Ferretti<sup>45,69</sup>, Adam Grundhoff<sup>401,407</sup>, Murat Iskar<sup>408</sup>, Kortine Kleinheinz<sup>52,54</sup>, Peter Lichter<sup>408</sup>, **Hidewaki Nakagawa**<sup>#48</sup>, Akinyemi I Ojesina<sup>255,256,257</sup>, Chandra Sekhar Pedamallu<sup>3,6,172</sup>, Matthias Schlesner<sup>52,110</sup>, Xiaoping Su<sup>144</sup> and Marc Zapatka<sup>408</sup>

### Tumor Specific Providers – Australia (Ovarian cancer)

Kathryn Alsop<sup>409,410</sup>, Australian Ovarian Cancer Study Group<sup>187,311,411</sup>, **David D L Bowtell**<sup>#187,291</sup>, Timothy JC Bruxner<sup>185</sup>, Angelika N Christ<sup>185</sup>, Elizabeth L Christie<sup>187</sup>, Stephen M Cordner<sup>412</sup>, Prue A Cowin<sup>187</sup>, Ronny Drapkin<sup>413</sup>, Dariush Etemadmoghadam<sup>187,188</sup>, Sian Fereday<sup>414</sup>, Dale W Garsed<sup>187,188</sup>, Joshy George<sup>170</sup>, Sean M Grimmond<sup>363</sup>, Anne Hamilton<sup>187</sup>, Oliver Holmes<sup>311,312</sup>, Jillian A Hung<sup>415,416</sup>, Karin S Kassahn<sup>185,417</sup>, Stephen H Kazakoff<sup>311,312</sup>, Catherine J Kennedy<sup>418,419</sup>, Conrad R Leonard<sup>311,312</sup>, Linda Mileskin<sup>187</sup>, David K Miller<sup>185,360,420</sup>, Gisela Mir Arnau<sup>187</sup>, Chris Mitchell<sup>187</sup>, Felicity Newell<sup>311,312</sup>,

Katia Nones<sup>311,312</sup>, Ann-Marie Patch<sup>311,312</sup>, John V Pearson<sup>311,312</sup>, Michael C Quinn<sup>311,312</sup>, Mark Shackleton<sup>188,218</sup>, Darrin F Taylor<sup>185</sup>, Heather Thorne<sup>187</sup>, Nadia Traficante<sup>187</sup>, Ravikiran Vedururu<sup>187</sup>, Nick M Waddell<sup>312</sup>, Nicola Waddell<sup>311,312</sup>, Paul M Waring<sup>253</sup>, Scott Wood<sup>311,312</sup>, Qinying Xu<sup>311,312</sup> and Anna deFazio<sup>421,422,423</sup>

### **Tumor Specific Providers – Australia (Pancreatic cancer)**

Matthew J Anderson<sup>185</sup>, Davide Antonello<sup>424</sup>, Andrew P Barbour<sup>425,426</sup>, Claudio Bassi<sup>424</sup>, Samantha Bersani<sup>427</sup>, **Andrew V Biankin**<sup>#359,360,361,362</sup>, Timothy JC Bruxner<sup>185</sup>, Ivana Cataldo<sup>427,428</sup>, David K Chang<sup>360,362</sup>, Lorraine A Chantrill<sup>360</sup>, Yoke-Eng Chiew<sup>421</sup>, Angela Chou<sup>360,429</sup>, Angelika N Christ<sup>185</sup>, Sara Cingarlini<sup>37</sup>, Nicole Cloonan<sup>430</sup>, Vincenzo Corbo<sup>428,431, 432</sup>, Fraser R Duthie<sup>433,434</sup>, J Lynn Fink<sup>105,185</sup>, Anthony J Gill<sup>360,435</sup>, Janet S Graham<sup>362,436</sup>, **Sean M Grimmond**<sup>#363</sup>, Ivon Harliwong<sup>185</sup>, Oliver Holmes<sup>311,312</sup>, Nigel B Jamieson<sup>361,362,437</sup>, Amber L Johns<sup>360,420</sup>, Karin S Kassahn<sup>185,417</sup>, Stephen H Kazakoff<sup>311,312</sup>, James G Kench<sup>360,435,438</sup>, Luca Landoni<sup>424</sup>, Rita T Lawlor<sup>428</sup>, Conrad R Leonard<sup>311,312</sup>, Andrea Mafficini<sup>428</sup>, Neil D Merrett<sup>424,439</sup>, David K Miller<sup>185,360,420</sup>, Marco Miotto<sup>424</sup>, Elizabeth A Musgrove<sup>362</sup>, Adnan M Nagrial<sup>360</sup>, Felicity Newell<sup>311,312</sup>, Katia Nones<sup>311,312</sup>, Karin A Oien<sup>253,440</sup>, Marina Pajic<sup>360</sup>, Ann-Marie Patch<sup>311,312</sup>, John V Pearson<sup>311,312</sup>, Mark Pinese<sup>360</sup>, Andreia V Pinho<sup>360</sup>, Michael C Quinn<sup>311,312</sup>, Alan J Robertson<sup>185</sup>, Ilse Rooman<sup>360</sup>, Borislav C Rusev<sup>428</sup>, Jaswinder S Samra<sup>424,435</sup>, Maria Scardoni<sup>427</sup>, Christopher J Scarlett<sup>360,441</sup>, Aldo Scarpa<sup>428</sup>, Elisabetta Sereni<sup>424</sup>, Katarzyna O Sikora<sup>428</sup>, Michele Simbolo<sup>431</sup>, Morgan L Taschuk<sup>45</sup>, Christopher W Toon<sup>360</sup>, Giampaolo Tortora<sup>37,38</sup>, Caterina Vicentini<sup>428</sup>, Nick M Waddell<sup>312</sup>, Nicola Waddell<sup>311,312</sup>, Scott Wood<sup>311,312</sup>, Jianmin Wu<sup>360</sup>, Qinying Xu<sup>311,312</sup> and Nikolajs Zeps<sup>442</sup>

### **Tumor Specific Providers – Australia (Skin cancer)**

Lauri A Aaltonen<sup>443</sup>, Andreas Behren<sup>444</sup>, Hazel Burke<sup>445</sup>, Jonathan Cebon<sup>444</sup>, Rebecca A Dagg<sup>446</sup>, Ricardo De Paoli-Iseppi<sup>447</sup>, Ken Dutton-Regester<sup>311</sup>, Matthew A Field<sup>448</sup>, Anna Fitzgerald<sup>449</sup>, Sean M Grimmond<sup>363</sup>, **Nicholas K Hayward**<sup>#311,445</sup>, Peter Hersey<sup>445</sup>, Oliver Holmes<sup>311,312</sup>, Valerie Jakrot<sup>445</sup>, Peter A Johansson<sup>311</sup>, Hojabr Kakavand<sup>447</sup>, Stephen H Kazakoff<sup>311,312</sup>, Richard F Kefford<sup>450</sup>, Loretta MS Lau<sup>451</sup>, Conrad R Leonard<sup>311,312</sup>, Georgina V Long<sup>452</sup>, **Graham J Mann**<sup>#453,454</sup>, Felicity Newell<sup>311,312</sup>, Katia Nones<sup>311,312</sup>, Ann-Marie Patch<sup>311,312</sup>, John V Pearson<sup>311,312</sup>, Hilda A Pickett<sup>451</sup>, Antonia L Pritchard<sup>311</sup>, Gulietta M Pupo<sup>455</sup>, Robyn PM Saw<sup>452</sup>, Sarah-Jane Schramm<sup>456</sup>, **Richard A Scolyer**<sup>#422,452,457,458</sup>, Mark Shackleton<sup>188,218</sup>, Catherine A Shang<sup>459</sup>, Ping Shang<sup>452</sup>, Andrew J Spillane<sup>452</sup>, Jonathan R Stretch<sup>452</sup>, Varsha Tembe<sup>456</sup>, John F Thompson<sup>452</sup>, Ricardo E Vilain<sup>457</sup>, Nick M Waddell<sup>312</sup>, Nicola Waddell<sup>311,312</sup>, James S Wilmott<sup>452</sup>, Scott Wood<sup>311,312</sup>, Qinying Xu<sup>311,312</sup> and Jean Y Yang<sup>460</sup>

### **Tumor Specific Providers – Canada (Pancreatic cancer)**

John Bartlett<sup>461,462</sup>, Prashant Bavi<sup>463</sup>, Ivan Boroza<sup>9</sup>, Dianne E Chadwick<sup>464</sup>, Michelle Chan-Seng-Yue<sup>463</sup>, Sean Cleary<sup>463,465</sup>, Ashton A Connor<sup>466,467</sup>, Karolina Czajka<sup>468</sup>, Robert E Denroche<sup>463</sup>, Neesha C Dhani<sup>469</sup>, Jenna Eagles<sup>79</sup>, Vincent Ferretti<sup>45,69</sup>, Steven Gallinger<sup>463,466,467</sup>, Robert C Grant<sup>463,470</sup>, David Hedley<sup>469</sup>, Michael A Hollingsworth<sup>471</sup>, **Thomas J Hudson**<sup>#78,79</sup>, Gun Ho Jang<sup>463</sup>, Jeremy Johns<sup>79</sup>, Sangeetha Kalimuthu<sup>463</sup>, Sheng-Ben Liang<sup>472</sup>, Ilinca Lungu<sup>463,473</sup>, Xuemei Luo<sup>9</sup>, Faridah Mbabaali<sup>79</sup>, **John D McPherson**<sup>#79,463,474</sup>, Treasa A McPherson<sup>470</sup>, Jessica K Miller<sup>79</sup>, Malcolm J Moore<sup>469</sup>, Faiyaz Notta<sup>463,475</sup>, Danielle Pasternack<sup>79</sup>, Gloria M Petersen<sup>476</sup>, Michael H A Roehrl<sup>133,463,477,478,479</sup>, Michelle

Sam<sup>79</sup>, Iris Selander<sup>470</sup>, Stefano Serra<sup>253</sup>, Sagedeh Shahabi<sup>472</sup>, **Lincoln D Stein**<sup>#9,10</sup>, Morgan L Taschuk<sup>45</sup>, Sarah P Thayer<sup>106</sup>, Lee E Timms<sup>79</sup>, Gavin W Wilson<sup>9,463</sup>, Julie M Wilson<sup>463</sup> and Bradly G Wouters<sup>480</sup>

### Tumor Specific Providers – Canada (Prostate cancer)

Timothy A Beck<sup>45</sup>, Vinayak Bhandari<sup>9</sup>, Paul C Boutros<sup>9,133,142,143</sup>, **Robert G Bristow**<sup>#133,481,482,483,484</sup>, Colin C Collins<sup>151</sup>, Shadrielle MG Espiritu<sup>9</sup>, Neil E Fleshner<sup>485</sup>, Natalie S Fox<sup>9</sup>, Michael Fraser<sup>9</sup>, Syed Haider<sup>9</sup>, Lawrence E Heisler<sup>486</sup>, Vincent Huang<sup>9</sup>, Emilie Lalonde<sup>9</sup>, Julie Livingstone<sup>9</sup>, John D McPherson<sup>79,463,474</sup>, Alice Meng<sup>487</sup>, Veronica Y Sabelnykova<sup>9</sup>, Adriana Salcedo<sup>9</sup>, Yu-Jia Shiah<sup>9</sup>, Theodorus Van der Kwast<sup>488</sup> and Takafumi N Yamaguchi<sup>9</sup>

### Tumor Specific Providers – China (Gastric cancer)

Shuai Ding<sup>489</sup>, Daiming Fan<sup>490</sup>, Yong Hou<sup>39,249</sup>, Yi Huang<sup>153,154</sup>, Lin Li<sup>39</sup>, Siliang Li<sup>39,249</sup>, Dongbing Liu<sup>39,249</sup>, Xingmin Liu<sup>39,249</sup>, **Youyong Lu**<sup>#28,29,30</sup>, Yongzhan Nie<sup>490,491</sup>, Hong Su<sup>39,249</sup>, Jian Wang<sup>39</sup>, Kui Wu<sup>39,249</sup>, Xiao Xiao<sup>154</sup>, Rui Xing<sup>29,492</sup>, **Huanming Yang**<sup>#39</sup>, Shanlin Yang<sup>489</sup>, Yingyan Yu<sup>493</sup>, <sup>230</sup>, Xiuqing Zhang<sup>39</sup>, Yong Zhou<sup>39</sup> and Shida Zhu<sup>39,249</sup>

### Tumor Specific Providers – EU: France (Renal cancer)

Rosamonde E Banks<sup>494</sup>, Guillaume Bourque<sup>495,496</sup>, Alvis Brazma<sup>7</sup>, Paul Brennan<sup>497</sup>, **Mark Lathrop**<sup>#496</sup>, Louis Letourneau<sup>498</sup>, Yasser Riazalhosseini<sup>496</sup>, Ghislaine Scelo<sup>497</sup>, **Jörg Tost**<sup>#499</sup>, Naveen Vasudev<sup>500</sup> and Juris Viksna<sup>501</sup>

### Tumor Specific Providers – EU: United Kingdom (Breast cancer)

Sung-Min Ahn<sup>502</sup>, Ludmil B Alexandrov<sup>2,317</sup>, Samuel Aparicio<sup>503</sup>, Laurent Arnould<sup>504</sup>, MR Aure<sup>505</sup>, Shriram G Bhosle<sup>2</sup>, E Birney<sup>7</sup>, Ake Borg<sup>506</sup>, S Boyault<sup>507</sup>, AB Brinkman<sup>508</sup>, JE Brock<sup>509</sup>, A Broeks<sup>510</sup>, Adam P Butler<sup>2</sup>, AL Børresen-Dale<sup>505</sup>, C Caldas<sup>511,512</sup>, Peter J Campbell<sup>1,2</sup>, Suet-Feung Chin<sup>511,512</sup>, Helen Davies<sup>2</sup>, C Desmedt<sup>513</sup>, L Dirix<sup>514</sup>, S Dronov<sup>2</sup>, Anna Ehinger<sup>515</sup>, JE Eyfjord<sup>516</sup>, GG Van den Eynden<sup>517</sup>, A Fatima<sup>217</sup>, Jorge Reis Filho<sup>518</sup>, JA Foekens<sup>519</sup>, PA Futreal<sup>520</sup>, Øystein Garred<sup>521,522</sup>, Moritz Gerstung<sup>7,8</sup>, Dilip D Giri<sup>518</sup>, D Glodzik<sup>2</sup>, Dorte Grabau<sup>523</sup>, Holmfridur Hilmarsdottir<sup>516</sup>, GK Hooijer<sup>524</sup>, Jocelyne Jacquemier<sup>525</sup>, SJ Jang<sup>526</sup>, Jon G Jonasson<sup>516</sup>, Jos Jonkers<sup>527</sup>, HY Kim<sup>525</sup>, Tari A King<sup>528,529</sup>, Stian Knappskog<sup>2</sup>, G Kong<sup>525</sup>, S Krishnamurthy<sup>530</sup>, S Van Laere<sup>514</sup>, SR Lakhani<sup>531</sup>, A Langerød<sup>505</sup>, Denis Larsimont<sup>532</sup>, HJ Lee<sup>526</sup>, JY Lee<sup>533</sup>, Ming Ta Michael Lee<sup>520</sup>, Yilong Li<sup>2</sup>, Ole Christian Lingjærde<sup>534</sup>, Gaetan MacGrogan<sup>535</sup>, JW Martens<sup>536</sup>, Sancha Martin<sup>2,369</sup>, Iñigo Martincorena<sup>2</sup>, Andrew Menzies<sup>2</sup>, Sandro Morganella<sup>2</sup>, Ville Mustonen<sup>320,321,322</sup>, Serena Nik-Zainal<sup>2,324,325,326</sup>, Sarah O'Meara<sup>2</sup>, I Pauporté<sup>18</sup>, Sarah Pinder<sup>537</sup>, X Pivot<sup>538</sup>, Elena Provenzano<sup>539</sup>, CA Purdie<sup>540</sup>, Keiran M Raine<sup>2</sup>, M Ramakrishna<sup>2</sup>, K Ramakrishnan<sup>2</sup>, AL Richardson<sup>217</sup>, M Ringnér<sup>506</sup>, Javier Bartolomé Rodríguez<sup>105</sup>, FG Rodríguez-González<sup>175</sup>, G Romieu<sup>541</sup>, Roberto Salgado<sup>253</sup>, Torill Sauer<sup>534</sup>, R Shepherd<sup>2</sup>, AM Sieuwerts<sup>177</sup>, PT Simpson<sup>531</sup>, M Smid<sup>542</sup>, C Sotiriou<sup>55</sup>, PN Span<sup>543</sup>, J Staaf<sup>506</sup>, Lucy Stebbings<sup>2</sup>, Ólafur Andri Stefánsson<sup>544</sup>, Alasdair Stenhouse<sup>545</sup>, **Michael Rudolf Stratton**<sup>#2</sup>, HG Stunnenberg<sup>249,546</sup>, Fred Sweep<sup>547</sup>, BK Tan<sup>548</sup>, Jon W Teague<sup>2</sup>, Gilles Thomas<sup>549</sup>, AM Thompson<sup>545</sup>, S Tommasi<sup>550</sup>, I Treilleux<sup>551,552</sup>, Andrew Tutt<sup>217</sup>, NT Ueno<sup>393</sup>, Peter Van Loo<sup>63,64</sup>, P Vermeulen<sup>514</sup>, Alain Viari<sup>428</sup>, MJ van de Vijver<sup>253</sup>, A Vincent-Salomon<sup>546</sup>,



David C Wedge<sup>2,354,355</sup>, Bernice Huimin Wong<sup>553</sup>, Lucy Yates<sup>2</sup>, X Zou<sup>2</sup>, CHM van Deurzen<sup>536</sup> and L van't Veer<sup>554,555</sup>

### **Tumor Specific Providers – Germany (Malignant lymphoma)**

Ole Ammerpohl<sup>556,557</sup>, Sietse Aukema<sup>558,559</sup>, Anke K Bergmann<sup>560</sup>, Stephan H Bernhart<sup>276,277,281</sup>, Hans Binder<sup>276,277</sup>, Arndt Borkhardt<sup>561</sup>, Christoph Borst<sup>562</sup>, Benedikt Brors<sup>82,119,278</sup>, Birgit Burkhardt<sup>563</sup>, Alexander Claviez<sup>564</sup>, Roland Eils<sup>52,54,66,67</sup>, Maria Elisabeth Goebler<sup>565</sup>, Andrea Haake<sup>556</sup>, Siegfried Haas<sup>562</sup>, Martin Hansmann<sup>566</sup>, Jessica I Hoell<sup>561</sup>, Steve Hoffmann<sup>277,279,280,281</sup>, Michael Hummel<sup>567</sup>, Daniel Hübschmann<sup>54,66,83,84,85</sup>, Dennis Karsch<sup>568</sup>, Wolfram Klapper<sup>559</sup>, Kortine Kleinheinz<sup>52,54</sup>, Michael Kneba<sup>568</sup>, Jan O Korbelt<sup>7,8</sup>, Helene Kretzmer<sup>277,281</sup>, Markus Kreuz<sup>569</sup>, Dieter Kube<sup>570</sup>, Ralf Küppers<sup>571</sup>, Chris Lawerenz<sup>67</sup>, Dido Lenze<sup>567</sup>, Peter Lichter<sup>408</sup>, Markus Loeffler<sup>569</sup>, Cristina López<sup>262,556</sup>, Luisa Mantovani-Löffler<sup>572</sup>, Peter Möller<sup>573</sup>, German Ott<sup>574</sup>, Bernhard Radlwimmer<sup>408</sup>, Julia Richter<sup>556,559</sup>, Marius Rohde<sup>575</sup>, Philip C Rosenstiel<sup>576</sup>, Andreas Rosenwald<sup>577</sup>, Markus B Schilhabel<sup>576</sup>, Matthias Schlesner<sup>52,110</sup>, Stefan Schreiber<sup>578</sup>, **Reiner Siebert**<sup>#262,761</sup>, Peter F Stadler<sup>276,277,281</sup>, Peter Staib<sup>579</sup>, Stephan Stilgenbauer<sup>580</sup>, Stephanie Sungalee<sup>8</sup>, Monika Szczepanowski<sup>559</sup>, Umut H Toprak<sup>54,581</sup>, Lorenz HP Trümper<sup>570</sup>, Rabea Wagener<sup>262,556</sup> and Thorsten Zenz<sup>82</sup>

### **Tumor Specific Providers – Germany (Pediatric Brain cancer)**

Ivo Buchhalter<sup>52,53,54</sup>, Juergen Eils<sup>66,67</sup>, Roland Eils<sup>52,54,66,67</sup>, Volker Hovestadt<sup>408</sup>, Barbara Hutter<sup>80,81,82</sup>, David TW Jones<sup>301,302</sup>, Natalie Jäger<sup>52</sup>, Christof von Kalle<sup>84</sup>, Marcel Kool<sup>98,301</sup>, Jan O Korbelt<sup>7,8</sup>, Andrey Korshunov<sup>98</sup>, Pablo Landgraf<sup>582</sup>, Chris Lawerenz<sup>67</sup>, Hans Lehrach<sup>583</sup>, **Peter Lichter**<sup>#408</sup>, Paul A Northcott<sup>584</sup>, Stefan M Pfister<sup>98,301,585</sup>, Bernhard Radlwimmer<sup>408</sup>, Guido Reifenberger<sup>582</sup>, Matthias Schlesner<sup>52,110</sup>, Hans-Jörg Warnatz<sup>583</sup>, Joachim Weischenfeldt<sup>8,113,114</sup>, Stephan Wolf<sup>586</sup>, Marie-Laure Yaspo<sup>583</sup> and Marc Zapatka<sup>408</sup>

### **Tumor Specific Providers – Germany (Prostate cancer)**

Yassen Assenov<sup>587</sup>, Benedikt Brors<sup>82,119,278</sup>, Juergen Eils<sup>66,67</sup>, Roland Eils<sup>52,54,66,67</sup>, Lars Feuerbach<sup>119</sup>, Clarissa Gerhauser<sup>285</sup>, Jan O Korbelt<sup>7,8</sup>, Chris Lawerenz<sup>67</sup>, Hans Lehrach<sup>583</sup>, Sarah Minner<sup>588</sup>, Christoph Plass<sup>285</sup>, **Guido Sauter**<sup>#589</sup>, Thorsten Schlomm<sup>114,590</sup>, Nikos Sidiropoulos<sup>113</sup>, Ronald Simon<sup>589</sup>, **Holger Sültmann**<sup>#82,591</sup>, Hans-Jörg Warnatz<sup>583</sup>, Dieter Weichenhan<sup>285</sup>, Joachim Weischenfeldt<sup>8,113,114</sup> and Marie-Laure Yaspo<sup>583</sup>

### **Tumor Specific Providers – India (Oral cancer)**

Nidhan K Biswas<sup>592</sup>, Luca Landoni<sup>424</sup>, Arindam Maitra<sup>592</sup>, **Partha P Majumder**<sup>#592</sup> and **Rajiv Sarin**<sup>#593</sup>

### **Tumor Specific Providers – Italy (Pancreatic cancer)**

Davide Antonello<sup>424</sup>, Stefano Barbi<sup>431</sup>, Claudio Bassi<sup>424</sup>, Samantha Bersani<sup>427</sup>, Giada Bonizzato<sup>428</sup>, Cinzia Cantù<sup>428</sup>, Ivana Cataldo<sup>427,428</sup>, Sara Cingarlini<sup>37</sup>, Vincenzo Corbo<sup>428,431, 432</sup>, Angelo P Dei Tos<sup>594</sup>, Matteo

Fassan<sup>595</sup>, Sonia Grimaldi<sup>428</sup>, Luca Landoni<sup>424</sup>, Rita T Lawlor<sup>428</sup>, Claudio Luchini<sup>427</sup>, Andrea Mafficini<sup>428</sup>, Giuseppe Malleo<sup>424</sup>, Giovanni Marchegiani<sup>424</sup>, Michele Milella<sup>37</sup>, Marco Miotto<sup>424</sup>, Salvatore Paiella<sup>424</sup>, Antonio Pea<sup>424</sup>, Paolo Pederzoli<sup>424</sup>, Borislav C Rusev<sup>428</sup>, Andrea Ruzzenente<sup>424</sup>, Roberto Salvia<sup>424</sup>, Maria Scardoni<sup>427</sup>, **Aldo Scarpa**<sup>#428</sup>, Elisabetta Sereni<sup>424</sup>, Michele Simbolo<sup>431</sup>, Nicola Sperandio<sup>428</sup>, Giampaolo Tortora<sup>37,38</sup> and Caterina Vicentini<sup>428</sup>

### **Tumor Specific Providers – Japan (Biliary tract cancer)**

Yasuhito Arai<sup>33</sup>, Natsuko Hama<sup>33</sup>, Nobuyoshi Hiraoka<sup>596</sup>, Fumie Hosoda<sup>33,597</sup>, Mamoru Kato<sup>366</sup>, Hiromi Nakamura<sup>33</sup>, Hidenori Ojima<sup>598</sup>, Takuji Okusaka<sup>599</sup>, **Tatsuhiko Shibata**<sup>#33,34</sup>, Yasushi Totoki<sup>33</sup> and Tomoko Urushidate<sup>34</sup>

### **Tumor Specific Providers – Japan (Gastric cancer)**

**Hiroyuki Aburatani**<sup>#272</sup>, Yasuhito Arai<sup>33</sup>, Masashi Fukayama<sup>600</sup>, Natsuko Hama<sup>33</sup>, Fumie Hosoda<sup>33,597</sup>, Shumpei Ishikawa<sup>601</sup>, Hitoshi Katai<sup>602</sup>, Mamoru Kato<sup>366</sup>, Hiroto Katoh<sup>603</sup>, Daisuke Komura<sup>601</sup>, Genta Nagae<sup>272,284</sup>, Hiromi Nakamura<sup>33</sup>, Hirofumi Rokutan<sup>604</sup>, Mihoko Saito-Adachi<sup>33</sup>, **Tatsuhiko Shibata**<sup>#33,34</sup>, Akihiro Suzuki<sup>272,605</sup>, Hirokazu Taniguchi<sup>606</sup>, Kenji Tatsuno<sup>272</sup>, Yasushi Totoki<sup>33</sup>, Tetsuo Ushiku<sup>600</sup>, Shinichi Yachida<sup>33,607</sup> and Shogo Yamamoto<sup>272</sup>

### **Tumor Specific Providers – Japan (Liver cancer)**

Hiroyuki Aburatani<sup>272</sup>, Hiroshi Aikata<sup>608</sup>, Koji Arihiro<sup>608</sup>, Shun-ichi Ariizumi<sup>609</sup>, Keith A Boroevich<sup>47,48</sup>, Kazuaki Chayama<sup>608</sup>, Akihiro Fujimoto<sup>48</sup>, Masashi Fujita<sup>48</sup>, Mayuko Furuta<sup>48</sup>, Kunihito Gotoh<sup>610</sup>, Natsuko Hama<sup>33</sup>, Takanori Hasegawa<sup>87</sup>, Shinya Hayami<sup>611</sup>, Shuto Hayashi<sup>87</sup>, Satoshi Hirano<sup>612</sup>, Seiya Imoto<sup>86,87</sup>, Mamoru Kato<sup>366</sup>, Yoshiiku Kawakami<sup>608</sup>, Kazuhiro Maejima<sup>48</sup>, Satoru Miyano<sup>87</sup>, Genta Nagae<sup>272,284</sup>, **Hidewaki Nakagawa**<sup>#48</sup>, Hiromi Nakamura<sup>33</sup>, Toru Nakamura<sup>612</sup>, Kaoru Nakano<sup>48</sup>, Hideki Ohdan<sup>608</sup>, Aya Sasaki-Oku<sup>48</sup>, **Tatsuhiko Shibata**<sup>#33,34</sup>, Yuichi Shiraishi<sup>87</sup>, Hiroko Tanaka<sup>87</sup>, Yasushi Totoki<sup>33</sup>, Tatsuhiko Tsunoda<sup>47,220,221,222</sup>, Masaki Ueno<sup>611</sup>, Rui Yamaguchi<sup>87</sup>, Masakazu Yamamoto<sup>609</sup> and Hiroki Yamaue<sup>611</sup>

### **Tumor Specific Providers – Singapore (Biliary tract cancer)**

Su Pin Choo<sup>613</sup>, Ioana Cutcutache<sup>267,319</sup>, Narong Khuntikeo<sup>424,614</sup>, John R McPherson<sup>267,319</sup>, Choon Kiat Ong<sup>615</sup>, Chawalit Pairajkul<sup>253</sup>, Irinel Popescu<sup>616</sup>, **Steven G Rozen**<sup>#267,268,319</sup>, **Patrick Tan**<sup>#254,266,267,268</sup> and **Bin Tean Teh**<sup>#266,267,268,269,270</sup>

### **Tumor Specific Providers – South Korea (Blood cancer)**

Keun Soo Ahn<sup>617</sup>, Hyung-Lae Kim<sup>27</sup>, Youngil Koh<sup>307,308</sup> and **Sung-Soo Yoon**<sup>#308</sup>

### **Tumor Specific Providers – Spain (Chronic Lymphocytic Leukemia)**

Marta Aymerich<sup>618</sup>, **Elias Campo**<sup>#619,620</sup>, Josep Ll Gelpi<sup>46,71</sup>, Ivo G Gut<sup>135,136</sup>, Marta Gut<sup>135,136</sup>, Armando Lopez-Guillermo<sup>621</sup>, Carlos López-Otín<sup>622</sup>, Xose S Puente<sup>623</sup>, Romina Royo<sup>105</sup> and David Torrents<sup>105,111</sup>

### **Tumor Specific Providers – United Kingdom (Bone cancer)**

Fernanda Amary<sup>624</sup>, Daniel Baumhoer<sup>625</sup>, Sam Behjati<sup>2</sup>, Bodil Bjerkehagen<sup>626</sup>, **Peter J Campbell**<sup>#1,2</sup>, **Adrienne M Flanagan**<sup>#627</sup>, PA Futreal<sup>520</sup>, Ola Myklebost<sup>628</sup>, Nischalan Pillay<sup>629</sup>, Patrick Tarpey<sup>630</sup>, Roberto Tirabosco<sup>631</sup> and Olga Zaikova<sup>632</sup>

### **Tumor Specific Providers – United Kingdom (Chronic myeloid disorders)**

Jacqueline Boulton<sup>633</sup>, David T Bowen<sup>2</sup>, Adam P Butler<sup>2</sup>, **Peter J Campbell**<sup>#1,2</sup>, Mario Cazzola<sup>634</sup>, Carlo Gambacorti-Passerini<sup>186</sup>, Anthony R Green<sup>295</sup>, Eva Hellstrom-Lindberg<sup>635</sup>, Luca Malcovati<sup>634</sup>, Sancha Martin<sup>2,369</sup>, Jyoti Nangalia<sup>636</sup>, Elli Papaemmanuil<sup>2</sup> and Paresh Vyas<sup>311,637</sup>

### **Tumor Specific Providers – United Kingdom (Esophageal cancer)**

Yeng Ang<sup>638</sup>, Hugh Barr<sup>639</sup>, Duncan Beardsmore<sup>640</sup>, Matthew Eldridge<sup>294</sup>, **Rebecca C Fitzgerald**<sup>#325</sup>, James Gossage<sup>641</sup>, Nicola Grehan<sup>325</sup>, George B Hanna<sup>642</sup>, Stephen J Hayes<sup>643,644</sup>, Ted R Hupp<sup>645</sup>, David Khoo<sup>646</sup>, Jesper Lagergren<sup>635,647</sup>, Laurence E Lovat<sup>251</sup>, Shona MacRae<sup>395</sup>, Maria O'Donovan<sup>325</sup>, J Robert O'Neill<sup>648</sup>, Simon L Parsons<sup>649</sup>, Shaun R Preston<sup>650</sup>, Sonia Puig<sup>651</sup>, Tom Roques<sup>652</sup>, Grant Sanders<sup>248</sup>, Sharmila Sothi<sup>653</sup>, Simon Tavaré<sup>294</sup>, Olga Tucker<sup>654</sup>, Richard Turkington<sup>655</sup>, Timothy J Underwood<sup>656</sup> and Ian Welch<sup>657</sup>

### **Tumor Specific Providers – United Kingdom (Prostate cancer)**

Nicholas Van As<sup>658</sup>, Daniel M Berney<sup>659</sup>, Johann S De Bono<sup>405</sup>, G Steven Bova<sup>330</sup>, Daniel S Brewer<sup>403,404</sup>, Adam P Butler<sup>2</sup>, Declan Cahill<sup>658</sup>, Niedzica Camacho<sup>405</sup>, **Colin S Cooper**<sup>#404,405,406</sup>, Nening M Dennis<sup>658</sup>, Tim Dudderidge<sup>658</sup>, Sandra E Edwards<sup>405</sup>, **Rosalind A Eeles**<sup>#405,658</sup>, Cyril Fisher<sup>658</sup>, Christopher S Foster<sup>660,661</sup>, Mohammed Ghori<sup>2</sup>, Pelvender Gill<sup>637</sup>, Vincent J Gnanapragasam<sup>383,662</sup>, Gunes Gundem<sup>2</sup>, Freddie C Hamdy<sup>663</sup>, Steve Hawkins<sup>294</sup>, Steven Hazell<sup>658</sup>, William Howat<sup>383</sup>, William B Isaacs<sup>292</sup>, Katalin Karaszi<sup>637</sup>, Jonathan D Kay<sup>251</sup>, Vincent Khoo<sup>658</sup>, Zsafia Kote-Jarai<sup>405</sup>, Barbara Kremeyer<sup>2</sup>, Pardeep Kumar<sup>658</sup>, Adam Lambert<sup>637</sup>, Daniel A Leongamornlert<sup>2,405</sup>, Naomi Livni<sup>658</sup>, Hayley J Luxton<sup>251</sup>, Andy G Lynch<sup>294,295,310</sup>, Luke Marsden<sup>637</sup>, Charlie E Massie<sup>294</sup>, Lucy Matthews<sup>405</sup>, Erik Mayer<sup>658,664</sup>, Ultan McDermott<sup>2</sup>, Sue Merson<sup>405</sup>, Thomas J Mitchell<sup>2,295,383</sup>, David E Neal<sup>294,383</sup>, Anthony Ng<sup>665</sup>, David Nicol<sup>658</sup>, Christopher Ogden<sup>658</sup>, Edward W Rowe<sup>658</sup>, Nimish C Shah<sup>383</sup>, Jon W Teague<sup>2</sup>, Sarah Thomas<sup>658</sup>, Alan Thompson<sup>658</sup>, Peter Van Loo<sup>63,64</sup>, Clare Verrill<sup>637,666</sup>, Tapio Visakorpi<sup>330</sup>, Anne Y Warren<sup>383,667</sup>, David C Wedge<sup>2,354,355</sup>, Hayley C Whitaker<sup>251</sup>, Yong-Jie Yu<sup>659</sup>, Yongwei Yu<sup>230</sup>, Jorge Zamora<sup>2,287,288,289</sup> and Hongwei Zhang<sup>230</sup>

### **Tumor Specific Providers – United States (TCGA)**

Adam Abeshouse<sup>199</sup>, Nishant Agrawal<sup>72</sup>, Rehan Akbani<sup>325,668</sup>, Hikmat Al-Ahmadie<sup>199</sup>, Monique Albert<sup>462</sup>, Kenneth Aldape<sup>253,646,669</sup>, Adrian Ally<sup>670</sup>, Yeng Ang<sup>638</sup>, Elizabeth L Appelbaum<sup>139,251</sup>, Joshua Armenia<sup>671</sup>, Sylvia Asa<sup>649,672</sup>, J Todd Auman<sup>673</sup>, Matthew H Bailey<sup>138,139</sup>, Miruna Balasundaram<sup>670</sup>, Saianand Balu<sup>248</sup>, Jill Barnholtz-Sloan<sup>674,675</sup>, Hugh Barr<sup>639</sup>, John Bartlett<sup>461,462</sup>, Oliver F Bathe<sup>676,677</sup>, Stephen B Baylin<sup>656,678</sup>, Duncan Beardsmore<sup>640</sup>, Christopher Benz<sup>679</sup>, Andrew Berchuck<sup>680</sup>, Benjamin P Berman<sup>273,274,275</sup>, Rameen Beroukhi<sup>3,6,172</sup>, Mario Berrios<sup>681</sup>, Darell Bigner<sup>294,682</sup>, Michael Birrer<sup>106</sup>, Tom Bodenheimer<sup>248</sup>, Lori Boice<sup>651</sup>, Moiz S Bootwalla<sup>683</sup>, Marcus Bosenberg<sup>684</sup>, Reanne Bowlby<sup>670</sup>, Jeffrey Boyd<sup>685</sup>, Russell R Broaddus<sup>669</sup>, Malcolm Brock<sup>686</sup>, Denise Brooks<sup>670</sup>, Susan Bullman<sup>3,172</sup>, Samantha J Caesar-Johnson<sup>40</sup>, Thomas E Carey<sup>687</sup>, Rebecca Carlsen<sup>670</sup>, Robert Cerfolio<sup>688</sup>, Vishal S Chandan<sup>689</sup>, Hsiao-Wei Chen<sup>638,671</sup>, Andrew D Cherniack<sup>3,3,49,172</sup>, Jeremy Chien<sup>690</sup>, Juok Cho<sup>3</sup>, Eric Chuah<sup>670</sup>, Carrie Cibulskis<sup>3</sup>, Kristian Cibulskis<sup>3</sup>, Leslie Cope<sup>691</sup>, Matthew G Cordes<sup>139,652</sup>, Kyle Covington<sup>165</sup>, Erin Curley<sup>692</sup>, Bogdan Czerniak<sup>646,669</sup>, Ludmila Danilova<sup>691</sup>, Ian J Davis<sup>693</sup>, Timothy Defreitas<sup>3</sup>, John A Demchok<sup>40</sup>, Noreen Dhalla<sup>670</sup>, Rajiv Dhir<sup>694</sup>, Li Ding<sup>138,139,146</sup>, HarshaVardhan Doddapaneni<sup>165</sup>, Adel El-Naggar<sup>646,669</sup>, Ina Felau<sup>40</sup>, Martin L Ferguson<sup>695</sup>, Gaetano Finocchiaro<sup>696</sup>, Kwun M Fong<sup>697</sup>, Scott Frazer<sup>3</sup>, William Friedman<sup>698</sup>, Catrina C Fronick<sup>139,652</sup>, Lucinda A Fulton<sup>139</sup>, Robert S Fulton<sup>138,139,146</sup>, Stacey B Gabriel<sup>3</sup>, Jianjiong Gao<sup>671</sup>, Nils Gehlenborg<sup>3,699</sup>, Jeffrey E Gershenwald<sup>700,701</sup>, Gad Getz<sup>3,4,5,6</sup>, Ronald Ghossein<sup>518</sup>, Nasra H Giama<sup>702</sup>, Richard A Gibbs<sup>165</sup>, Carmen Gomez<sup>703</sup>, James Gossage<sup>641</sup>, Ramaswamy Govindan<sup>138</sup>, Nicola Grehan<sup>325</sup>, George B Hanna<sup>642</sup>, D Neil Hayes<sup>248,704,705</sup>, Stephen J Hayes<sup>643,644</sup>, Apurva M Hegde<sup>395,668</sup>, David I Heiman<sup>3</sup>, Zachary Heins<sup>199</sup>, Austin J Hepperla<sup>248</sup>, Katherine A Hoadley<sup>247,248</sup>, Andrea Holbrook<sup>706</sup>, Robert A 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